

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property  
Organization  
International Bureau



(43) International Publication Date  
3 June 2004 (03.06.2004)

PCT

(10) International Publication Number  
**WO 2004/046388 A1**

(51) International Patent Classification<sup>7</sup>: C12Q 001/68

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(21) International Application Number:

PCT/AU2003/001544

(81) Designated States (*national*): AE, AG, AL, AM, AT, AU,  
AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR,  
CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD,  
GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR,  
KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN,  
MW, MX, MZ, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU,  
SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA,  
UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.

(22) International Filing Date:

18 November 2003 (18.11.2003)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

2002952702 18 November 2002 (18.11.2002) AU

(84) Designated States (*regional*): ARIPO patent (BW, GH,  
GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW),  
Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM),  
European patent (AT, BE, BG, CH, CY, CZ, DE, DK, EE,  
ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO, SE,  
SI, SK, TR), OAPI patent (BF, BJ, CF, CG, CI, CM, GA,  
GN, GQ, GW, ML, MR, NE, SN, TD, TG).

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Published:

— with international search report

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*For two-letter codes and other abbreviations, refer to the "Guid-  
ance Notes on Codes and Abbreviations" appearing at the begin-  
ning of each regular issue of the PCT Gazette.*

(54) Title: GENOTYPING OF DEAFNESS BY OLIGONUCLEOTIDE MICROARRAY ANALYSIS

(57) Abstract: A method for genotyping a subject with respect to a gene or target nucleic acid sequence associated with a patho-  
logical condition, said method comprising contacting an allele specific oligonucleotide immobilized to a solid support with a sin-  
gle-stranded form of RNA or DNA from a subject to be tested labeled directly or indirectly with a reporter molecule capable of giving  
an identifiable signal under conditions which permit hybridization of single stranded RNA or DNA which is exactly complementary  
to the immobilized allele specific oligonucleotide but substantially less or no hybridization of non-complementary single-stranded  
RNA or DNA molecules and then screening for the presence or absence or level of reporter molecule which provides an indicator of  
the genetic identity of the single-stranded RNA or DNA molecule which in turn provides the genotype of the subject.

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## GENOTYPING OF DEAFNESS BY OLIGONUCLEOTIDE MICROARRAY ANALYSIS

### BACKGROUND OF THE INVENTION

#### 5. FIELD OF THE INVENTION

The present invention relates generally to a method for genotyping a subject to identify a likelihood of that subject developing a pathological condition. More particularly, the present invention provides genotyping of deafness or an associated disorder using  
10 hybridization of single-stranded testRNA or DNA to a sequence-specific oligonucleotide. Even more particularly, the present invention employs microarray analysis to identify the presence of heterozygous or homozygous wild-type or mutant sequences of a gene or other nucleic acid target. This provides the genotype of a particular gene or nucleic acid target. The present invention may be provided in kit form and may be conducted manually,  
15 automatically or semi-automatically. The identification of a subject's genotype with respect to a gene or other target nucleic acid facilitates corrective therapy at the medical or behavioral level.

#### DESCRIPTION OF THE PRIOR ART

20

Bibliographic details of references provided in the subject specification are listed at the end of the specification.

Reference to any prior art in this specification is not, and should not be taken as, an  
25 acknowledgment or any form of suggestion that this prior art forms part of the common general knowledge in any country.

Deafness is one of the most common human genetic conditions. Approximately one child in 1000 is born with a prelingual hearing loss which will have a significant impact on the  
30 infant's speech, language and general development, incurring lifelong social, educational and economic costs (Yoshinaga, *Otolaryngol Clin. North Am.* 32(6): 1089-1102, 199?).

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Approximately 10% of the population are affected by age-related hearing loss by the age of 60 years and 50% by the age of 80 years (Davis, Hearing in adults, London: Whurr, 1995). More than half of prelingual deafness has a genetic basis and defects in many genes, probably more than 100, can cause deafness. More than 20 genes have been identified to date (Petit *et al.*, *Annu. Rev. Genet.* 35: 589-646, 2001). Despite this genetic heterogeneity, a small group of genes are known to account for the majority of genetic non-syndromic hearing loss. For example, mutations in the *connexin 26* gene are responsible for over half of autosomal recessive non-syndromic hearing loss. Mutations in the *pendrin* gene can cause both non-syndromic and syndromic (Pendred Syndrome) deafness and are estimated to cause up to 10% of genetic hearing loss. The A1555G mitochondrial 12S rRNA mutation has been reported at a high frequency in Spanish and Japanese families with severe progressive deafness and can induce hearing loss upon exposure to aminoglycosides, which are commonly given in high doses to premature babies. Mutations in the *usherin* gene are largely responsible for the most common form of Usher Syndrome, type II, which is characterized by congenital deafness with onset of retinitis pigmentosa in late teens (Van Camp and Smith, Hereditary hearing loss homepage, URL: <http://dnalab-www.uia.ac.be/dnalab/hhh/>).

The genetic heterogeneity of deafness has proved a challenge for genetic testing: analysis of multiple genes by conventional gel-based methods is both time-consuming and expensive. There is a need, therefore, to develop more efficient and accurate means of identifying mutations or polymorphisms in genes and nucleic acid molecules associated with genetic deafness.

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## SUMMARY OF THE INVENTION

Throughout this specification, unless the context requires otherwise, the word "comprise", or variations such as "comprises" or "comprising", will be understood to imply the  
5 inclusion of a stated element or integer or group of elements or integers but not the exclusion of any other element or integer or group of elements or integers.

Nucleotide and amino acid sequences are referred to by a sequence identifier number (SEQ ID NO:). The SEQ ID NOs: correspond numerically to the sequence identifiers <400>1  
10 (SEQ ID NO:1), <400>2 (SEQ ID NO:2), etc. A summary of the sequence identifiers is provided in Table 1. A sequence listing is provided at the end of the specification.

The present invention is directed to a sequence-specific oligonucleotide-based genotyping of one or more target genes or target nucleic acid molecules in a single subject or in  
15 multiple subjects. More particularly, the present invention employs sequence-specific oligonucleotides directed to particular alleles or mutations or polymorphisms in genes or other nucleic acid molecules (e.g. rRNA) associated with a pathological condition such as deafness. Genetic deafness is heterogenous and there are more than 60 linked loci and more than 20 genes associated with this condition. The present invention combines  
20 microarray technology with sequence specific oligonucleotide hybridization to screen for one or a multiplicity of genes in a single subject or in a number of subjects. The sequence-specific oligonucleotide is also referred to herein as an allele-specific oligonucleotide.

The nucleic acid microarray, or biochip, is a new hybridization-based genotyping  
25 technique that offers simultaneous analysis of many genetic mutations. The parallelism offered by the microarray platform makes it ideally suited to genotyping of genetically heterogeneous conditions such as deafness.

Allele-specific oligonucleotides to genes or other target nucleic acid molecule such as  
30 *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* are immobilized onto a solid support. The solid support is preferably planar such as on a microchip or biochip.

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However, the present invention is also applicable on spheres and nanoparticles, each coded by a reporter molecule or other characteristic feature. RNA or DNA from a subject to be tested is amplified and labeled with a reporter molecule and rendered single-stranded before being brought into contact with the immobilized allele-specific oligonucleotides.

5

Alternatively, the presence or absence of a test RNA or DNA which has hybridized to an immobilized sequence specific oligonucleotide may be achieved by hybridizing a labeled oligonucleotide (referred to as a reporter oligonucleotide) to, for example, a particular nucleotide sequence on the target RNA or DNA distinct from the nucleotide sequence  
10 which encompasses the mutation. Conveniently, a nucleotide tail of, for example, Ts or As may be used as a generic tag for a reporter oligonucleotide.

Still in a further alternative, the label may be a nucleotide capable of creating a current. Such nucleotides are referred to as an *electrotide*. Such technology uses the complementary  
15 binding properties of RNA or DNA to create an electric circuit.

Hybridization or non-hybridization is determined by the presence or absence of the signal of the reporter molecule. An algorithm is then used to define the genotype index (GI), wherein:

20

$$GI = \frac{SV_N}{SV_N + SV_M}$$

wherein:

25  $SV_N$  is the normal spot value; and  
 $SV_M$  is the mutant spot value.

The value is the level of signal of the reporter molecule. Preferably, the reporter molecule is a fluorescent molecule including a fluorophore.

30

- 5 -

The present invention provides, therefore, a method for genotyping a subject with respect to a gene or target nucleic acid sequence associated with a pathological condition, said method comprising contacting an allele specific oligonucleotide immobilized to a solid support with a single-stranded form of RNA or DNA from a subject to be tested labeled  
5 directly or indirectly with a reporter molecule capable of giving an identifiable signal under conditions which permit hybridization of single stranded RNA or DNA which is exactly complementary to the immobilized allele specific oligonucleotide but substantially less or no hybridization of non-complementary single-stranded RNA or DNA molecules and then screening for the presence or absence or level of reporter molecule which  
10 provides an indicator of the genetic identity of the single-stranded RNA or DNA molecule which in turn provides the genotype of the subject.

Examples of preferred oligonucleotides are shown in Table 1. The oligonucleotides may have a sequence of particular nucleotides or of a single type of nucleotide at the  
15 immobilization end of the molecule. This is the case for SEQ ID NOs:1 to 32 which have  $[T]_x$  where x is 10. Alternatively, a chemical linker may be used between the solid support and the oligonucleotide. Furthermore, the target sequence may be modified using mismatched primers to interrupt sequences of particular nucleotides which may otherwise adversely affect hybridization.

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A summary of the allele specific oligonucleotides and corresponding SEQ ID NOs is shown in Table 1 for each gene tested.

TABLE 1

*List of allele specific oligonucleotides and genes used to detect genotype*

GENE	MUTATION	OLIGONUCLEOTIDE SEQUENCE	SEQ ID NO.
<b>Connexin 26</b>			
35delG	35W18A	TTTTTTTTTTGATCCTGGGGGGTGTGAA	1
	35W18B	TTTTTTTTTTATCCTGGGGGGTGTGPAC	2
	35M17A	TTTTTTTTTTGATCCTGGGGGGTGTGAA	3
	35M17B	TTTTTTTTTTATCCTGGGGGGTGTGAAC	4
M34T	34W16	TTTTTTTTTTTCGCATTATGATCCTCG	5
	34M16	TTTTTTTTTTTCGCATTACGATCCTCG	6
167delT	167W18	TTTTTTTTTTCAACACCCTGCAGCCAGG	7
	167M17	TTTTTTTTTTCAACACCCGCAGCCAGG	8
235delC	235W16	TTTTTTTTTTTATGGGCCCTGCAGCT	9
	235M15	TTTTTTTTTTTATGGGCCCTGCAGCT	10
V37I	37W16	TTTTTTTTTTTGATCCTCGTTGTGGCT	11
	37M18	TTTTTTTTTTTGATCCTCATTGTGGCTG	12
W24X	24W19	TTTTTTTTTTTGAAAGATCTGGCTCACCGT	13
	24M19	TTTTTTTTTTTGAAAGATCTAGCTCACCGT	14
L90P	90W19	TTTTTTTTTTTCAGCGCTCCTAGTGCCAT	15
	90W17	TTTTTTTTTTTAGCGCTCCAGTGCCCA	16
R143W	143W17	TTTTTTTTTTTCTTCTCCGGGTCATC	17
	143M19	TTTTTTTTTTTATCTTCTTCTGGGTCATCT	18
313del14	313W20	TTTTTTTTTTTAGGAAGTTCATCAAGGGGA	19
	313M20	TTTTTTTTTTTGAAGAAGAGGGGAGATAAAG	20
<b>Pendrin</b>			
L236P	236W21	TTTTTTTTTTGTCTCACAGCTAAAGATTGTC	21
	236M21	TTTTTTTTTTGTCTCACAGCCAAAGATTGTC	22
100I	1001W17	TTTTTTTTTTTCCAAGCGGGTGAGTGTG	23
	1001M18	TTTTTTTTTTTCCAAGGGGATGAGTGTGG	24
E384G	384W21	TTTTTTTTTTGTCTCCTTAGGAATTCATTGCC	25
	384M19	TTTTTTTTTTTTCCTTAGGGATTCATTGC	26
T416P	416W19	TTTTTTTTTTTCAGGAGAGCACTGGAGGAA	27
	416M19	TTTTTTTTTTTCAGGAGAGCCCTGGAGGAA	28
<b>Mitochondrial rRNA</b>			
A1555G	1555W21	TTTTTTTTTTTATAGAGGAGACAAGTCGTAA	29

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GENE	MUTATION	OLIGONUCLEOTIDE SEQUENCE	SEQ ID NO.
	1555M21	TTTTTTTTTTTATAGAGGAGGCAAGTCGTAA	30
<b>Usher1n</b>			
2299delG	2299W20	TTTTTTTTTTGGGCAGTGTGAGTGCAAAAA	31
	2299M17	TTTTTTTTTTGGGCAGTGTAGTGCAAAA	32
<b>Connexin 26</b>			
35delG	35W18A	GATCCTGGGGGGTGTGAA	33
	35W18B	ATCCTGGGGGGTGTGAAC	34
	35M17A	GATCCTGGGGGGTGTGAA	35
	35M17B	ATCCTGGGGGGTGTGAAC	36
M34T	34W16	CGCATTATGATCCTCG	37
	34M16	CGCATTACGATCCTCG	38
167delT	167W18	CAACACCCTGCAGCCAGG	39
	167M17	CAACACCCGCAGCCAGG	40
235delC	235W16	ATGGGCCCTGCAGCT	41
	235M15	ATGGGCCCTGCAGCT	42
V37I	37W16	GATCCTCGTTGTGGCT	43
	37M18	GATCCTCATTGTGGCTG	44
W24X	24W19	GAAAGATCTGGCTCACCCT	45
	24M19	GAAAGATCTAGCTCACCCT	46
L90P	90W19	CAGCGCTCCTAGTGGCCAT	47
	90W17	AGCGCTCCCAGTGGCCA	48
R143W	143W17	CTTCTTCCGGGTCATC	49
	143M19	ATCTTCTTGTGGGTCATCT	50
313del14	313W20	AGGAAGTTCATCAAGGGGGA	51
	313M20	GAAGAAGAGGGGAGATAAAG	52
<b>Pendrin</b>			
L236P	236W21	GTCTCACAGCTAAAGATTGTC	53
	236M21	GTCTCACAGCCAAAGATTGTC	54
100I	1001W17	CCAAGGGGGTGAAGTGTG	55
	1001M18	CCAAGGGGATGAAGTGTGG	56
E384G	384W21	GCTTCCTTAGGAATTCATTGCC	57
	384M19	CCTTAGGGATTCATTGC	58
T416P	416W19	CAGGAGAGCACTGGAGGAA	59
	416M19	CAGGAGAGCCCTGGAGGAA	60
<b>Mitochondrial rRNA</b>			
A1555G	1555W21	ATAGAGGAGACAAGTCGTAA	61
	1555M21	ATAGAGGAGGCAAGTCGTAA	62
<b>Usher1n</b>			
2299delG	2299W20	GGGCAGTGTGAGTGCAAAAA	63
	2299M17	GGCAGTGTAGTGCAAAA	64

## BRIEF DESCRIPTION OF THE FIGURES

**Figure 1** is a diagrammatic representation showing attachment chemistry for allele-specific oligonucleotides to microarray solid support.

5

**Figure 2** is a diagrammatic representation showing microarray based genotyping using allele-specific oligonucleotides.

**Figure 3** is a photographic representation showing genotyping of *connexin 26* 35ΔG and M34T mutations.

10

**Figure 4** is a graphical representation of the genotype index (GI) of *connexin 26* 35ΔG and M34T mutations.

**Figure 5** is a photographic representation of genotyping of *connexin 26* mutations 35ΔG/M3RT, 35DG/35ΔG, M34T/M34T, 167delT/N, 167delT/167delT, 235delC/N and V371/N. N = normal; M = mutant.

15

**Figure 6** is a photographic representation of genotyping of *pendrin* and 12S rRNA mutations. *Pendrin*: 1001 G > A, E384G, T416P and L236P. 12SrRNA: A1555G. N = normal; M = mutant.

20

**Figures 7(a)-(n)** are graphical representations showing the genotype index (GI) of various genes associated with deafness.

25

**Figure 8** is a graphical representation of genotype algorithms to determine N/N (homozygous normal), N/M (heterozygous normal) and M/M (homozygous mutant).

**Figure 9** is a graphical and tabular representation showing interactions between deafness genes.

30

## DETAILED DESCRIPTION OF THE PREFERRED EMBODIMENTS

The present invention provides a method for genotyping a particular subject with respect to a gene or other target nucleic acid molecule such as an mRNA or rRNA. More particularly, the present invention combines allele (i.e. sequence) specific oligonucleotide hybridization specificity with microarray analysis in order to genotype a subject with respect to a gene or genes or other target nucleic acid molecules associated with a pathological condition.

Accordingly, one aspect of the present invention contemplates a method for genotyping a subject with respect to a gene or target nucleic acid sequence associated with a pathological condition, said method comprising contacting an allele specific oligonucleotide immobilized to a solid support with a single-stranded form of RNA or DNA from a subject to be tested labeled directly or indirectly with a reporter molecule capable of giving an identifiable signal under conditions which permit hybridization of single stranded RNA or DNA which is exactly complementary to the immobilized allele specific oligonucleotide but substantially less or no hybridization of non-complementary single-stranded RNA or DNA molecules and then screening for the presence or absence or level of reporter molecule which provides an indicator of the genetic identity of the single-stranded RNA or DNA molecule which in turn provides the genotype of the subject.

Reference to direct or indirect labeling includes incorporating a label or a labeled nucleotide into the test RNA or DNA during PCR or alternatively using labeled oligonucleotides which hybridize to portions of the test RNA or DNA not associated with a mutation. For example, a sequence of nucleotides such as As, T, Gs or Cs or mixtures thereof may be added to a target RNA or DNA. A labeled oligonucleotide sequence complementary to the introduced nucleotide sequence is then used to determine the presence or absence of an immobilized target RNA or DNA sequence.

A label includes a reporter molecule capable of giving an identifiable signal (e.g. a fluorescent molecule) or a nucleotide capable of creating an electrical current or other electrical signal.

- 10 -

The present invention applies to a range of pathological conditions within a range of subjects. Such subjects include humans, non-human primates, livestock animals, laboratory test animals, companion animals and captured wild animals.

5

Pathological conditions contemplated herein include but are not limited to myopathy, obesity, anorexia, weight maintenance, diabetes, disorders associated with mitochondrial dysfunction, genetic disorders, cancer, heart disease, inflammation, disorders associated with the immune system, (e.g. autoimmune disease), infertility, disease associated with the  
10 brain, neurological disorders and neurodegenerative disorders.

As used herein, "myopathy" refers to any abnormal conditions or disease of the muscle tissues, which include the muscles over our bones (skeletal muscle) and the heart (cardiac muscle).

15

Diseases and conditions contemplated by the present invention include Alzheimer's, Parkinson's, diabetes, autism, and the aging process, Lethal Infantile Cardio myopathy, Beta-oxidation Defects, COX Deficiency, Mitochondrial Cytopathy, Alpers Disease, Barth syndrome, Carnitine-Acyl-Carnitine Deficiency, Carnitine Deficiency, Co-Enzyme Q10  
20 Deficiency, Complex I Deficiency, Complex II Deficiency, Complex III Deficiency, Complex IV Deficiency, Complex V Deficiency, CPEO, CPT I Deficiency, Glutaric Aciduria Type II, KSS, lactic acidosis, LCAD, LCHAD, Leigh Disease, LHON, Luft Disease, MAD, MCA, MELAS, MERRF, mitochondrial DNA depletion, Mitochondrial Encephalopath, MNGIE, NARP, Pearson Syndrome, Pyruvate Carboxylase Deficiency,  
25 Pyruvate Dehydrogenase Deficiency, SCAD, SCHAD and VLCAD; Alpers Disease, or Progressive Infantile Poliodystrophy, includes symptoms such as seizures, dementia, spasticity, blindness, liver dysfunction, and cerebral degeneration; Barth syndrome is an X-linked recessive disorder the symptoms of which include skeletal myopathy, cardio myopathy, short stature, and neutropenia; Carnitine-Acyl-Carnitine Deficiency is an  
30 autosomal recessive disorder, the symptoms of which are seizures, apnea, bradycardia, vomiting, lethargy, coma, enlarged liver, limb weakness, myoglobin in the urine, Reye-like

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symptoms triggered by fasting; Carnitine Deficiency is an autosomal recessive disease, the  
 symptoms of which include Cardio myopathy, failure to thrive, and altered consciousness  
 or coma, sometimes hypotonia; A-Beta-Lipoproteinemia, A-V, A Beta-2-Microglobulin  
 Amyloidosis, A-T, AlAD, AlAT, Aagenaes, Aarskog syndrome, Aarskog-Scott  
 5 Syndrome, Aase-smith syndrome, Aase Syndrome, AAT, Abderhalden-Kaufmann-Lignac  
 Syndrome, Abdominal Muscle Deficiency Syndrome, Abdominal Wall Defect, Abdominal  
 Epilepsy, Abdominal Migraine, Abductor Spasmodic Dysphonia, Abductor Spastic  
 Dysphonia, Abercrombie Syndrome, blepharon-Macrostomia Syndrome, ABS, Absence of  
 HPRT, Absence of Corpus Callosum Schinzel Typ, Absence Defect of Limbs Scalp and  
 10 Skull, Absence of Menstruation Primar, Absence of HGPRT, Absorptive Hyperoxaluriaor  
 Enteric, Abt-Letterer-Siwe Disease, ACADL, ACADM Deficiency, ACADM, ACADS,  
 Acanthocytosis-Neurologic Disorder, Acanthocytosis, Acantholysis Bullosa, Acanthosis  
 Nigricans, Acanthosis Bullosa, Acanthosis Nigricans With Insulin Resistance Type A,  
 Acanthosis Nigricans With Insulin Resistance Type B, Acanthotic Nevus, Acatalasemia,  
 15 Acatalasia, ACC, Accessory Atrioventricular Pathways, Accessory Atrioventricular  
 Pathways, Acephaly, ACF with Cardiac Defects, Achalasia, Achard-Thiers Syndrome,  
 ACHARD (Marfan variant), Achard's syndrome, Acholuric Jaundice, Achondrogenesis,  
 Achondrogenesis Type IV, Achondrogenesis Type III, Achondroplasia, Achondroplasia  
 Tarda, Achondroplastic Dwarfism, Achoo Syndrome, Achromat, Achromatope,  
 20 Achromatopic, Achromatopsia, Achromic Nevi, Acid Ceramidase Deficiency, Acid  
 Maltase Deficiency, Acid Beta-glucosidase Deficiency, Acidemia Methylmalonic,  
 Acidemia Propionic, Acidemia with Episodic Ataxia and Weakness, Acidosis, Aclasis  
 Tarsoepiphyseal, ACM, Acoustic Neurilemoma, Acoustic Neuroma, ACPS with Leg  
 Hypoplasia, ACPS II, ACPS IV, ACPS III, Acquired Aphasia with Convulsive Disorder,  
 25 Acquired Brown Syndrome, Acquired Epileptic Aphasia, Acquired Factor XIII Deficiency,  
 Acquired Form of ACC (caused by infection while still in womb), Acquired  
 Hyperoxaluria, Acquired Hypogammaglobulinemia, Acquired Immunodeficiency  
 Syndrome (AIDS), Acquired Iron Overload, Acquired Lipodystrophy, Acquired Partial  
 Lipodystrophy, Acquired Wandering Spleen, ACR, Acral Dysostosis with Facial and  
 30 Genital Abnormalities, Acro Renal, Acrocallosal Syndrome Schinzel Type,  
 Acrocephalosyndactyly, Acrocephalosyndactyly Type I, Acrocephalosyndactyly Type I

Subtype I, Acrocephalopolysyndactyly Type II, Acrocephalopolysyndactyly Type III, Acrocephalopolysyndactyly Type IV, Acrocephalosyndactyly V (ACS5 or ACS V) Subtype I, Acrocephaly Skull Asymmetry and Mild Syndactyly, Acrocephaly, Acrochondrohyperplasia, Acrodermatitis Enteropathica, Acrodysostosis, Acrodystrophic  
5 Neuropathy, Acrofacial Dysostosis Nager Type, Acrofacial Dysostosis Postaxial Type, Acrofacial Dysostosis Type Genee-Wiedep, Acrogeria Familial, Acromegaly, Acromelalgia Hereditary, Acromesomelic Dysplasia, Acromesomelic Dwarfism, Acromicric Skeletal Dysplasia, Acromicric Dysplasia, Acroosteolysis with Osteoporosis and Changes in Skull and Mandible, Acroosteolysis, Acroparesthesia, ACS I, ACS Type  
10 II, ACS Type III, ACS, ACS3, ACTH Deficiency, Action Myoclonus, Acute Brachial Neuritis Syndrome, Acute Brachial Radiculitis Syndrome, Acute Cerebral Gaucher Disease, Acute Cholangitis, Acute Disseminated Encephalomyeloradiculopathy, Acute Disseminated Histiocytosis-X, Acute Hemorrhagic Polioencephalitis, Acute Idiopathic Polyneuritis, Acute Immune-Mediation Polyneuritis, Acute Infantile Pelizaeus-Merzbacher  
15 Brain Sclerosis, Acute Intermittant Porphyrin, Acute Porphyrin, Acute Sarcoidosis, Acute Shoulder Neuritis, Acute Toxic Epidermolysis, Acyl-CoA Dehydrogenase Deficiency Long-Chain, Acyl-CoA Dehydrogenase Deficiency Short-Chain, Acyl-CoA Dihydroxyacetone Acyltransferase, Acyl-coenzyme A Oxidase Deficiency, ADA, ADA Deficiency, Adam Complex, Adamantiades-Behcet's Syndrome, Adamantinoma, Adams  
20 Oliver Syndrome, Adaptive Colitis, ADD combined type, ADD, Addison Disease with Cerebral Sclerosis, Addison's Anemia, Addison's Disease, Addison-Biermer Anemia, Addison-Schilder Disease, Addisonian Pernicious Anemia, Adducted Thumbs-Mental Retardation, Adductor Spasmodic Dysphonia, Adductor Spastic Dysphonia, Adenoma Associated Virilism of Older Women, Adenomatosis of the Colon and Rectum,  
25 Adenomatous polyposis of the Colon, Adenomatous Polyposis Familial, Adenosine Deaminase Deficiency, Adenylosuccinase deficiency, ADHD predominantly hyperactive-impulsive type, ADHD predominantly inattentive type, ADHD, Adhesive Arachnoiditis, Adie Syndrome, Adie's Syndrome, Adie's Tonic Pupil, Adie's Pupil, Adipogenital Retinitis Pigmentosa Polydactyly, Adipogenital-Retinitis Pigmentosa Syndrome, Adiposa  
30 Dolorosa, Adiposis Dolorosa, Adiposogenital Dystrophy, Adolescent Cystinosis, ADPKD, Adrenal Cortex Adenoma, Adrenal Disease, Adrenal Hyperfunction resulting from

Pituitary ACTH Excess, Adrenal Hypoplasia, Adrenal Insufficiency, Adrenal Neoplasm,  
 Adrenal Virilism, Adreno-Retinitis Pigmentosa-Polydactyly Syndrome, Adrenocortical  
 Insufficiency, Adrenocortical Hypofunction, Adrenocorticotrophic Hormone Deficiency  
 Isolated, Adrenogenital Syndrome, Adrenoleukodystrophy, Adrenomyeloneuropathy,  
 5 Adreno-Retinitis Pigmentosa-Polydactyly Syndrome, Adult Cystinosis, Adult  
 Dermatomyositis, Adult Hypophosphatasia, Adult Macula Lutea Retinae Degeneration,  
 Adult Onset ALD, Adult-Onset Ceroidosis, Adult Onset Medullary Cystic Disease, Adult  
 Onset Pernicious Anemia, Adult Onset Schindler Disease, Adult-Onset Subacute  
 Necrotizing Encephalomyelopathy, Adult Polycystic Kidney Disease, Adult Onset  
 10 Medullary Cystic Disease, Adynlosuccinate Lyase Deficiency, AE, AEC Syndrome, AFD,  
 Afibrinogenemia, African Siderosis, AGA, Aganglionic Megacolon, Age Related Macular  
 Degeneration, Agenesis of Commissura Magna Cerebri, Agenesis of Corpus Callosum,  
 Agenesis of Corpus Callosum-Infantile Spasms-Ocular Anomalies, Agenesis of Corpus  
 Callosum and Chorioretinal Abnormality, Agenesis of Corpus Callosum-Chorioretinitis  
 15 Abnormality, Aggressive mastocytosis, Agnosis Primary, AGR Triad, AGU, Agyria,  
 Agyria-pachygyria-band spectrum, AHC, AHD, AHDS, AHF Deficiency, AHG Deficiency,  
 AHO, Ahumada Del Castillo, Aicardi Syndrome, AIED, AIMP, AIP, AIS, Akinetic  
 Seizure, ALA-D Porphyrria, Alactasia, Alagille Syndrome, Aland Island Eye Disease (X-  
 Linked), Alaninuria, Albers-Schonberg Disease, Albinism, Albinismus, Albinoidism,  
 20 Albright Hereditary Osteodystrophy, Alcaptonuria, Alcohol-Related Birth Defects,  
 Alcoholic Embryopathy, Ald, ALD, ALD, Aldosterone, Aldosteronism With Normal  
 Blood Pressure, Aldrich Syndrome, Alexander's Disease, Alexanders Disease,  
 Algodystrophy, Algoneurodystrophy, Alkaptonuria, Alkaptonuric Ochronosis, Alkyl  
 DHAP synthase deficiency, Allan-Herndon-Dudley Syndrome, Allan-Herndon Syndrome,  
 25 Allan-Herndon-Dudley Mental Retardation, Allergic Granulomatous Antitis, Allergic  
 Granulomatous Angiitis of Cronkhite-Canada, Alobar Holoprosencephaly, Alopecia  
 Areata, Alopecia Celsi, Alopecia Cicatrisata, Alopecia Circumscripta, Alopecia-Poliosis-  
 Uveitis-Vitiligo-Deafness-Cutaneous-Uveo-O, Alopecia Seminuniversalis, Alopecia  
 Totalis, Alopecia Universalis, Alpers Disease, Alpers Diffuse Degeneration of Cerebral  
 30 Gray Matter with Hepatic Cirrhosis, Alpers Progressive Infantile Poliodystrophy, Alpha-1-  
 Antitrypsin Deficiency, Alpha-1 4 Glucosidase Deficiency, Alpha-Galactosidase A

Deficiency, Alpha-Galactosidase B Deficiency, Alpha High-Density Lipoprotein  
 Deficiency, Alpha-L-Fucosidase Deficiency Fucosidosis Type 3, Alpha-GalNAc Deficiency  
 Schindler Type, Alpha-lipoproteinemia, Alpha Mannosidosis, Alpha-N-  
 Acetylgalactosaminidase Deficiency Schindler Type, Alpha-NAGA Deficiency Schindler  
 5 Type, Alpha-Neuraminidase Deficiency, Alpha-Thalassemia/mental retardation syndrome  
 non-deletion type, Alpha-lipoproteinemia, Alport Syndrome, ALS, Alstroem's Syndrome,  
 Alstroem, Alstrom Syndrome, Alternating Hemiplegia Syndrome, Alternating Hemiplegia  
 of Childhood, Alzheimer's Disease, Amaurotic Familial Idiocy, Amaurotic Familial Idiocy  
 Adult, Amaurotic Familial Infantile Idiocy, Ambiguous Genitalia, AMC, AMD,  
 10 Ameloblastoma, Amelogenesis Imperfecta, Amenorrhea-Galactorrhea Nonpuerperal,  
 Amenorrhea-Galactorrhea-FSH Decrease Syndrome, Amenorrhea, Amino Acid Disorders,  
 Aminoaciduria-Osteomalacia-Hyperphosphaturia Syndrome, AMN, Amniocentesis,  
 Amniotic Bands, Amniotic Band Syndrome, Amniotic Band Disruption Complex,  
 Amniotic Band Sequence, Amniotic Rupture Sequence, Amputation Congenital, AMS,  
 15 Amsterdam Dwarf Syndrome de Lange, Amylo-1 6-Glucosidase Deficiency, Amyloid  
 Arthropathy of Chronic Hemodialysis, Amyloid Corneal Dystrophy, Amyloid  
 Polyneuropathy, Amyloidosis, Amyloidosis of Familial Mediterranean Fever,  
 Amylopectinosis, Amyoplasia Congenita, Amyotrophic Lateral Sclerosis, Amyotrophic  
 Lateral Sclerosis, Amyotrophic Lateral Sclerosis-Polyglucosan Bodies, AN, AN 1, AN 2,  
 20 Anal Atresia, Anal Membrane, Anal Rectal Malformations, Anal Stenosis, Analine 60  
 Amyloidosis, Analphalipoproteinemia, Analrectal, Analrectal, Anaplastic Astrocytoma,  
 Andersen Disease, Anderson-Fabry Disease, Andersen Glycogenosis, Anderson-Warburg  
 Syndrome, Andre Syndrome, Andre Syndrome Type II, Androgen Insensitivity, Androgen  
 Insensitivity Syndrome Partial, Androgen Insensitivity Syndrome Partial, Androgenic  
 25 Steroids, Anemia Autoimmune Hemolytic, Anemia Blackfan Diamond, Anemia,  
 Congenital, Triphalangeal Thumb Syndrome, Anemia Hemolytic Cold Antibody, Anemia  
 Hemolytic with PGK Deficiency, Anemia Pernicious, Anencephaly, Angelman Syndrome,  
 Angio-Osteohypertrophy Syndrome, Angiofollicular Lymph Node Hyperplasia,  
 Angiohemophilia, Angiokeratoma Corporis, Angiokeratoma Corporis Diffusum,  
 30 Angiokeratoma Diffuse, Angiomatosis Retina, Angiomatous Lymphoid, Angioneurotic  
 Edema Hereditary, Anhidrotic Ectodermal Dysplasia, Anhidrotic X-Linked Ectodermal

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Dysplasias, Aniridia, Aniridia-Ambiguous Genitalia-Mental Retardation, Aniridia  
 Associated with Mental Retardation, Aniridia-Cerebellar Ataxia-Mental Deficiency,  
 Aniridia Partial-Cerebellar Ataxia-Mental Retardation, Aniridia Partial-Cerebellar Ataxia-  
 Oligophrenia, Aniridia Type I, Aniridia Type II, Aniridia-Wilms' Tumor Association,  
 5 Aniridia-Wilms' Tumor-Gonadoblastoma, Ankyloblepharon-Ectodermal Defects-Cleft  
 Lip/Palate, Ankylosing Spondylitis, Annular grooves, Anodontia, Anodontia Vera,  
 Anomalous Trichromasy, Anomalous Dysplasia of Dentin, Coronal Dentin Dysplasia,  
 Anomic Aphasia, Anophthalmia, Anorectal, Anorectal Malformations, Anosmia, Anterior  
 Bowing of the Legs with Dwarfism, Anterior Membrane Corneal Dystrophy, Anti-  
 10 Convulsant Syndrome, Anti-Epstein-Barr Virus Nuclear Antigen (EBNA) Antibody  
 Deficiency, Antibody Deficiency, Antibody Deficiency with near normal  
 Immunoglobulins, Antihemophilic Factor Deficiency, Antihemophilic Globulin  
 Deficiency, Antiphospholipid Syndrome, Antiphospholipid Antibody Syndrome,  
 Antithrombin III Deficiency, Antithrombin III Deficiency Classical (Type I), Antitrypsin  
 15 Deficiency, Antley-Bixler Syndrome, Antoni's Palsy, Anxietas Tibialis, Aorta Arch  
 Syndrome, Aortic and Mitral Atresia with Hypoplastic Left Heart Syndrome, Aortic  
 Stenosis, Apatoschisis, APC, APECED Syndrome, Apert Syndrome, Aperts, Aphasia,  
 Aplasia Axialis Extracorticales Congenital, Aplasia Cutis Congenita, Aplasia Cutis  
 Congenita with Terminal Transverse Limb Defects, Aplastic Anemia, Aplastic Anemia  
 20 with Congenital Anomalies, APLS, Apnea, Appalachian Type Amyloidosis, Apple Peel  
 Syndrome, Apraxia, Apraxia Buccofacial, Apraxia Constructional, Apraxia Ideational,  
 Apraxia Ideokinetic, Apraxia Ideomotor, Apraxia Motor, Apraxia Oculomotor, APS,  
 Arachnitis, Arachnodactyly Contractural Beals Type, Arachnodactyly, Arachnoid Cysts,  
 Arachnoiditis Ossificans, Arachnoiditis, Aran-Duchenne, Aran-Duchenne Muscular  
 25 Atrophy, Aregenerative Anemia, Arginase Deficiency, Argininemia, Arginino Succinase  
 Deficiency, Argininosuccinase Deficiency, Argininosuccinate Lyase Deficiency,  
 Argininosuccinic Acid Lyase-ASL, Argininosuccinic Acid Synthetase Deficiency,  
 Argininosuccinic Aciduria, Argonz-Del Castillo Syndrome, Arhinencephaly, Armenian  
 Syndrome, Arnold-Chiari Malformation, Arnold-Chiari Syndrome, ARPKD, Arrhythmic  
 30 Myoclonus, Arrhythmogenic Right Ventricular Dysplasia, Arteriohepatic Dysplasia,  
 Arteriovenous Malformation, Arteriovenous Malformation of the Brain, Arteritis Giant

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Cell, Arthritis, Arthritis Urethritica, Arthro-Dento-Osteodysplasia, Arthro-Ophthalmopathy, Arthrochhalasis Multiplex Congenita, Arthrogryposis Multiplex Congenita, Arthrogryposis Multiplex Congenita, Distal, Type IIA, ARVD, Arylsulfatase-B Deficiency, AS, ASA Deficiency, Ascending Paralysis, ASD, Atrioseptal Defects, ASH, 5 Ashermans Syndrome, Ashkenazi Type Amyloidosis, ASL Deficiency, Aspartylglucosaminuria, Aspartylglycosaminuria, Asperger's Syndrome, Asperger's Type Autism, Asphyxiating Thoracic Dysplasia, Asplenia Syndrome, ASS Deficiency, Asthma, Astrocytoma Grade I (Benign), Astrocytoma Grade II (Benign), Asymmetric Crying Facies with Cardiac Defects, Asymmetrical septal hypertrophy, Asymptomatic Callosal 10 Agenesis, AT, AT III Deficiency, AT III Variant IA, AT III Variant Ib, AT 3, Ataxia, Ataxia Telangiectasia, Ataxia with Lactic Acidosis Type II, Ataxia Cerebral Palsy, Ataxiodynamia, Ataxiophemia, ATD, Athetoid Cerebral Palsy, Atopic Eczema, Atresia of Esophagus with or without Tracheoesophageal Fistula, Atrial Septal Defects, Atrial Septal Defect Primum, Atrial and Septal and Small Ventricular Septal Defect, Atrial Flutter, 15 Atrial Fibrillation, Atriadigital Dysplasia, Atrioseptal Defects, Atrioventricular Block, Atrioventricular Canal Defect, Atrioventricular Septal Defect, Atrophia Bulborum Hereditaria, Atrophic Beriberi, Atrophy Olivopontocerebellar, Attention Deficit Disorder, Attention Deficit Hyperactivity Disorder, Attenuated Adenomatous Polyposis Coli, Atypical Amyloidosis, Atypical Hyperphenylalaninemia, Auditory Canal Atresia, 20 Auriculotemporal Syndrome, Autism, Autism Asperger's Type, Autism Dementia Ataxia and Loss of Purposeful Hand Use, Autism Infantile Autism, Autoimmune Addison's Disease, Autoimmune Hemolytic Anemia, Autoimmune Hepatitis, Autoimmune-Polyendocrinopathy-Candidias, Autoimmune Polyglandular Disease Type I, Autosomal Dominant Albinism, Autosomal Dominant Compelling Helioophthalmic Outburst 25 Syndrome, Autosomal Dominant Desmin Distal myopathy with Late Onset, Autosomal Dominant EDS, Autosomal Dominant Emery-Dreifuss Muscular Dystrophy, Autosomal Dominant Keratoconus, Autosomal Dominant Pelizaeus-Merzbacher Brain Sclerosis, Autosomal Dominant Polycystic Kidney Disease, Autosomal Dominant Spinocerebellar Degeneration, Autosomal Recessive Agammaglobulinemia, Autosomal Recessive 30 Centronuclear myopathy, Autosomal Recessive Conradi-Hunermann Syndrome, Autosomal Recessive EDS, Autosomal Recessive Emery-Dreifuss Muscular Dystrophy,

Autosomal Recessive Forms of Ocular Albinism, Autosomal Recessive Inheritance  
 Agenesis of Corpus Callosum, Autosomal Recessive Keratoconus, Autosomal Recessive  
 Polycystic Kidney Disease, Autosomal Recessive Severe Combined Immunodeficiency,  
 AV, AVM, AVSD, AWTa, Axilla Abscess, Axonal Neuropathy Giant, Azorean  
 5 Neurologic Disease, B-K Mole Syndrome, Babinski-Froelich Syndrome, BADS,  
 Baillarger's Syndrome, Balkan Disease, Baller-Gerold Syndrome, Ballooning Mitral  
 Valve, Balo Disease Concentric Sclerosis, Baltic Myoclonus Epilepsy, Bannayan-Zonana  
 syndrome (BZS), Bannayan-Riley-Ruvalcaba syndrome, Banti's Disease, Bardet-Biedl  
 Syndrome, Bare Lymphocyte Syndrome, Barlow's syndrome, Barraquer-Simons Disease,  
 10 Barrett Esophagus, Barrett Ulcer, Barth Syndrome, Bartter's Syndrome, Basal Cell Nevus  
 Syndrome, Basedow Disease, Bassen-Kornzweig Syndrome, Batten Disease, Batten-  
 Mayou Syndrome, Batten-Spielmeier-Vogt's Disease, Batten Turner Syndrome, Batten  
 Turner Type Congenital myopathy, Batten-Vogt Syndrome, BBB Syndrome, BBB  
 Syndrome (Opitz), BBB Syndrome, BBBG Syndrome, BCKD Deficiency, BD, BDLs, BE,  
 15 Beals Syndrome, Beals Syndrome, Beals-Hecht Syndrome, Bean Syndrome, BEB,  
 Bechterew Syndrome, Becker Disease, Becker Muscular Dystrophy, Becker Nevus,  
 Beckwith Wiedemann Syndrome, Beckwith-Syndrome, Begnez-Cesar's Syndrome,  
 Behcet's syndrome, Behcet's Disease, Behr 1, Behr 2, Bell's Palsy, Benign Acanthosis  
 Nigricans, Benign Astrocytoma, Benign Cranial Nerve Tumors, Benign Cystinosis, Benign  
 20 Essential Blepharospasm, Benign Essential Tremor, Benign Familial Hematuria, Benign  
 Focal Amyotrophy, Benign Focal Amyotrophy of ALS, Benign Hydrocephalus, Benign  
 Hypermobility Syndrome, Benign Keratosis Nigricans, Benign Paroxysmal Peritonitis,  
 Benign Recurrent Hematuria, Benign Recurrent Intrahepatic Cholestasis, Benign Spinal  
 Muscular Atrophy with Hypertrophy of the Calves, Benign Symmetrical Lipomatosis,  
 25 Benign Tumors of the Central Nervous System, Berardinelli-Seip Syndrome, Berger's  
 Disease, Beriberi, Berman Syndrome, Bernard-Horner Syndrome, Bernard-Soulier  
 Syndrome, Besnier Prurigo, Best Disease, Beta-Alanine-Pyruvate Aminotransferase, Beta-  
 Galactosidase Deficiency Morquio Syndrome, Beta-Glucuronidase Deficiency, Beta  
 Oxidation Defects, Beta Thalassemia Major, Beta Thalassemia Minor, Betalipoprotein  
 30 Deficiency, Bethlehem myopathy, Beuren Syndrome, BH4 Deficiency, Biber-Haab-Dimmer  
 Corneal Dystrophy, Bicuspid Aortic Valve, Biedl-Bardet, Bifid Cranium, Bifunctional

- Enzyme Deficiency, Bilateral Acoustic Neurofibromatosis, Bilateral Acoustic Neuroma, Bilateral Right-Sidedness Sequence, Bilateral Renal Agenesis, Bilateral Temporal Lobe Disorder, Bilious Attacks, Bilirubin Glucuronosyltransferase Deficiency Type I, Binder Syndrome, Binswanger's Disease, Binswanger's Encephalopathy, Biotinidase deficiency,
- 5 Bird-Headed Dwarfism Seckel Type, Birth Defects, Birthmark, Bitemporal Forceps Marks Syndrome, Biventricular Fibrosis, Bjornstad Syndrome, B-K Mole Syndrome, Black Locks-Albinism-Deafness of Sensoneural Type (BADS), Blackfan-Diamond Anemia, Bleennorrheal Idiopathic Arthritis, Blepharophimosis, Ptosis, Epicanthus Inversus Syndrome, Blepharospasm, Blepharospasm Benign Essential, Blepharospasm
- 10 Oromandibular Dystonia, Blessig Cysts, BLFS, Blindness, Bloch-Siemens Incontinentia Pigmenti Melanoblastosis Cutis Linearis, Bloch-Siemens-Sulzberger Syndrome, Bloch-Sulzberger Syndrome, Blood types, Blood type A, Blood type B, Blood type AB, Blood type O, Bloom Syndrome, Bloom-Torre-Mackacek Syndrome, Blue Rubber Bleb Nevus, Blue Baby, Blue Diaper Syndrome, BMD, BOD, BOFS, Bone Tumor-Epidermoid Cyst-
- 15 Polyposis, Bonnet-Dechaume-Blanc Syndrome, Bonnevie-Ulrich Syndrome, Book Syndrome, BOR Syndrome, BORJ, Borjeson Syndrome, Borjeson-Forssman-Lehmann Syndrome, Bowen Syndrome, Bowen-Conradi Syndrome, Bowen-Conradi Hutterite, Bowen-Conradi Type Hutterite Syndrome, Bowman's Layer, BPEI, BPES, Brachial Neuritis, Brachial Neuritis Syndrome, Brachial Plexus Neuritis, Brachial-Plexus-
- 20 Neuropathy, Brachiocephalic Ischemia, Brachmann-de Lange Syndrome, Brachycephaly, Brachymorphic Type Congenital, Bradycardia, Brain Tumors, Brain Tumors Benign, Brain Tumors Malignant, Branched Chain Alpha-Ketoacid Dehydrogenase Deficiency, Branched Chain Ketonuria I, Brancher Deficiency, Branchio-Oculo-Facial Syndrome, Branchio-Oto-Renal Dysplasia, Branchio-Oto-Renal Syndrome, Branchiooculofacial Syndrome,
- 25 Branchiootic Syndrome, Brandt Syndrome, Brandywine Type Dentinogenesis Imperfecta, Brandywine type Dentinogenesis Imperfecta, Breast Cancer, BRIC Syndrome, Brittle Bone Disease, Broad Beta Disease, Broad Thumb Syndrome, Broad Thumbs and Great Toes Characteristic Facies and Mental Retardation, Broad Thumb-Hallux, Broca's Aphasia, Brocq-Duhring Disease, Bronze Diabetes, Bronze Schilder's Disease, Brown
- 30 Albinism, Brown Enamel Hereditary, Brown-Sequard Syndrome, Brown Syndrome, BRRS, Brueghel Syndrome, Bruton's Agammaglobulinemia Common, BS, BSS,

Buchanan's Syndrome, Budd's Syndrome, Budd-Chiari Syndrome, Buerger-Gruetz Syndrome, Bulbospinal Muscular Atrophy-X-linked, Bulldog Syndrome, Bullosa Hereditaria, Bullous CIE, Bullous Congenital Ichthyosiform Erythroderma, Bullous Ichthyosis, Bullous Pemphigoid, Burkitt's Lymphoma, Burkitt's Lymphoma African type, 5 Burkitt's Lymphoma Non-african type, BWS, Byler's Disease, C Syndrome, C1 Esterase Inhibitor Dysfunction Type II Angioedema, C1-INH, C1 Esterase Inhibitor Deficiency Type I Angioedema, C1NH, Cacchi-Ricci Disease, CAD, CADASIL, CAH, Calcaneal Valgus, Calcaneovalgus, Calcium Pyrophosphate Dihydrate Deposits, Callosal Agenesis and Ocular Abnormalities, Calves-Hypertrophy of Spinal Muscular Atrophy, Campomelic 10 Dysplasia, Campomelic Dwarfism, Campomelic Syndrome, Camptodactyly-Cleft Palate-Clubfoot, Camptodactyly-Limited Jaw Excursion, Camptomelic Dwarfism, Camptomelic Syndrome, Camptomelic Syndrome Long-Limb Type, Camurati-Engelmann Disease, Canada-Cronkhite Disease, Canavan disease, Canavan's Disease Included, Canavan's Leukodystrophy, Cancer, Cancer Family Syndrome Lynch Type, Cantrell Syndrome, 15 Cantrell-Haller-Ravich Syndrome, Cantrell Pentalogy, Carbamyl Phosphate Synthetase Deficiency, Carbohydrate Deficient Glycoprotein Syndrome, Carbohydrate-Deficient Glycoprotein Syndrome Type Ia, Carbohydrate-Induced Hyperlipemia, Carbohydrate Intolerance of Glucose Galactose, Carbon Dioxide Acidosis, Carboxylase Deficiency Multiple, Cardiac-Limb Syndrome, Cardio-auditory Syndrome, Cardioauditory Syndrome 20 of Jervell and Lange-Nielsen, Cardiocutaneous Syndrome, Cardio-facial-cutaneous syndrome, Cardiofacial Syndrome Cayler Type, Cardiomegalia Glycogenica Diffusa, Cardiomyopathic Lentiginosis, Cardio myopathy, Cardio myopathy Associated with Desmin Storage myopathy, Cardio myopathy Due to Desmin Defect, Cardio myopathy-Neutropenia Syndrome, Cardio myopathy-Neutropenia Syndrome Lethal Infantile Cardio 25 myopathy, Cardiopathic Amyloidosis, Cardiospasm, Cardocardiac Syndrome, Carnitine-Acylcarnitine Translocase Deficiency, Carnitine Deficiency and Disorders, Carnitine Deficiency Primary, Carnitine Deficiency Secondary, Carnitine Deficiency Secondary to MCAD Deficiency, Carnitine Deficiency Syndrome, Carnitine Palmitoyl Transferase I & II (CPT I & II), Carnitine Palmitoyltransferase Deficiency, Carnitine Palmitoyltransferase 30 Deficiency Type 1, Carnitine Palmitoyltransferase Deficiency Type 2 benign classical muscular form included severe infantile form included, Carnitine Transport Defect

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(Primary Carnitine Deficiency), Carnosinase Deficiency, Carnosinemia, Caroli Disease, Carpenter syndrome, Carpenter's, Cartilage-Hair Hypoplasia, Castleman's Disease, Castleman's Disease Hyaline Vascular Type, Castleman's Disease Plasma Cell Type, Castleman Tumor, Cat Eye Syndrome, Cat's Cry Syndrome, Catalayse deficiency,

5 Cataract-Dental Syndrome, Cataract X-Linked with Hutchinsonian Teeth, Catecholamine hormones, Catel-Manzke Syndrome, Catel-Manzke Type Palatodigital Syndrome, Caudal Dysplasia, Caudal Dysplasia Sequence, Caudal Regression Syndrome, Causalgia Syndrome Major, Cavernomas, Cavernous Angioma, Cavernous Hemangioma, Cavernous Lymphangioma, Cavernous Malformations, Cayler Syndrome, Cazenave's Vitiligo,

10 CBGD, CBPS, CCA, CCD, CCHS, CCM Syndrome, CCMS, CCO, CD, CDG1a, CDG1A, CDGS Type Ia, CDGS, CDI, CdLS, Celiac Disease, Celiac sprue, Celiac Sprue-Dermatitis, Cellular Immunodeficiency with Purine Nucleoside Phosphorylase Deficiency, Celsus' Vitiligo, Central Apnea, Central Core Disease, Central Diabetes Insipidus, Central Form Neurofibromatosis, Central Hypoventilation, Central Sleep Apnea, Centrifugal

15 Lipodystrophy, Centronuclear myopathy, CEP, Cephalocele, Cephalothoracic Lipodystrophy, Ceramide Trihexosidase Deficiency, Cerebellar Agenesis, Cerebellar Aplasia, Cerebellar Hemiagenesis, Cerebellar Hypoplasia, Cerebellar Vermis Aplasia, Cerebellar Vermis Agenesis-Hypernea-Episodic Eye Moves-Ataxia-Retardation, Cerebellar Syndrome, Cerebellarparenchymal Disorder IV, Cerebellomedullary

20 Malformation Syndrome, Cerebello-Oculocutaneous Telangiectasia, Cerebelloparenchymal Disorder IV Familial, Cerebellopontine Angle Tumor, Cerebral Arachnoiditis, Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukodystrophy, Cerebral Beriberi, Cerebral Diplegia, Cerebral Gigantism, Cerebral Malformations Vascular, Cerebral Palsy, Cerebro-Oculorenal Dystrophy, Cerebro-Oculo-

25 Facio-Skeletal Syndrome, Cerebrocostomandibular syndrome, Cerebrohepatorenal Syndrome, Cerebromacular Degeneration, Cerebromuscular Dystrophy Fukuyama Type, Cerebroocular Dysgenesis, Cerebroocular Dysplasia-Muscular Dystrophy Syndrome, Cerebrooculofacioskeletal Syndrome, Cerebroretinal Arteriovenous Aneurysm, Cerebroside Lipidosis, Cerebrosidosis, Cerebrotendinous Xanthomatosis, Cerebrovascular

30 Ferrocalcinosis, Ceroid-Lipofuscinosis Adult form, Cervical Dystonia, Cervical Dystonia, Cervico-Oculo-Acoustic Syndrome, Cervical Spinal Stenosis, Cervical Vertebral Fusion,

CES, CF, CFC syndrome, CFIDS, CFND, CGD, CGF, Chalasoderma Generalized,  
 Chanarin Dorfman Disease, Chanarin Dorfman Syndrome, Chanarin Dorfman Ichthyosis  
 Syndrome, Chandler's Syndrome, Charcot's Disease, Charcot-Marie-Tooth, Charcot-  
 Marie-Tooth Disease, Charcot-Marie-Tooth Disease Variant, Charcot-Marie-Tooth-  
 5 Roussy-Levy Disease, CHARGE Association, Charge Syndrome, CHARGE Syndrome,  
 Chaund's Ectodermal Dysplasias, Chediak-Higashi Syndrome, Chediak-Steinbrinck-  
 Higashi Syndrome, Cheilitis Granulomatosa, Cheiloschisis, Chemke Syndrome, Cheney  
 Syndrome, Cherry Red Spot and Myoclonus Syndrome, CHF, CHH, Chiari's Disease,  
 Chiari Malformation I, Chiari Malformation, Chiari Type I (Chiari Malformation I), Chiari  
 10 Type II (Chiari Malformation II), Chiari I Syndrome, Chiari-Budd Syndrome, Chiari-  
 Frommel Syndrome, Chiari Malformation II, CHILD Syndrome, CHILD Ichthyosis  
 Syndrome, CHILD Syndrome Ichthyosis, Childhood Adrenoleukodystrophy, Childhood  
 Dermatomyositis, Childhood-onset Dystonia, Childhood Cyclic Vomiting, Childhood  
 Giant Axonal Neuropathy, Childhood Hypophosphatasia, Childhood Muscular Dystrophy,  
 15 CHN, Cholestasis, Cholestasis Hereditary Norwegian Type, Cholestasis Intrahepatic,  
 Cholestasis Neonatal, Cholestasis of Oral Contraceptive Users, Cholestasis with Peripheral  
 Pulmonary Stenosis, Cholestasis of Pregnancy, Cholesterol Desmolase Deficiency,  
 Chondrodysplasia Punctata, Chondrodystrophia Calcificans Congenita, Chondrodystrophia  
 Fetalis, Chondrodystrophic Myotonia, Chondrodystrophy, Chondrodystrophy with  
 20 Clubfeet, Chondrodystrophy Epiphyseal, Chondrodystrophy Hyperplastic Form,  
 Chondroectodermal Dysplasias, Chondrogenesis Imperfecta, Chondrohystrophia,  
 Chondroosteodystrophy, Choreoacanthocytosis, Chorionic Villi Sampling, Choriorretinal  
 Anomalies, Choriorretinal Anomalies with ACC, Chorireninal Coloboma-Joubert  
 Syndrome, Choroidal Sclerosis, Choroideremia, Chotzen Syndrome, Christ-Siemens-  
 25 Touraine Syndrome, Christ-Siemans-Touraine Syndrome, Christmas Disease, Christmas  
 Tree Syndrome, Chromosome 3 Deletion of Distal 3p, Chromosome 3 Distal 3p  
 Monosomy, Chromosome 3-Distal 3q2 Duplication, Chromosome 3-Distal 3q2 Trisomy,  
 Chromosome 3 Monosomy 3p2, Chromosome 3q Partial Duplication Syndrome,  
 Chromosome 3q, Partial Trisomy Syndrome, Chromosome 3-Trisomy 3q2, Chromosome 4  
 30 Deletion 4q31-qter Syndrome, Chromosome 4 Deletion 4q32-qter Syndrome,  
 Chromosome 4 Deletion 4q33-qter Syndrome, Chromosome 4 Long Arm Deletion,

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Chromosome 4 Long Arm Deletion, Chromosome 4 Monosomy 4q, Chromosome 4-  
 Monosomy 4q, Chromosome 4 Monosomy Distal 4q, Chromosome 4 Partial Deletion 4p,  
 Chromosome 4, Partial Deletion of the Short Arm, Chromosome 4 Partial Monosomy of  
 Distal 4q, Chromosome 4 Partial Monosomy 4p, Chromosome 4 Partial Trisomy 4 (q25-  
 5 qter), Chromosome 4 Partial Trisomy 4 (q26 or q27-qter), Chromosome 4 Partial Trisomy  
 4 (q31 or 32-qter), Chromosome 4 Partial Trisomy 4p, Chromosome 4 Partial Trisomies  
 4q2 and 4q3, Chromosome 4 Partial Trisomy Distal 4, Chromosome 4 Ring, Chromosome  
 4 4q Terminal Deletion Syndrome, Chromosome 4q- Syndrome, Chromosome 4q-  
 Syndrome, Chromosome 4 Trisomy 4, Chromosome 4 Trisomy 4p, Chromosome 4 XY/47  
 10 XXY (Mosaic), Chromosome 5 Monosomy 5p, Chromosome 5, Partial Deletion of the  
 Short Arm Syndrome, Chromosome 5 Trisomy 5p, Chromosome 5 Trisomy 5p Complete  
 (5p11-pter), Chromosome 5 Trisomy 5p Partial (5p13 or 14-pter), Chromosome 5p-  
 Syndrome, Chromosome 6 Partial Trisomy 6q, Chromosome 6 Ring, Chromosome 6  
 Trisomy 6q2, Chromosome 7 Monosomy 7p2, Chromosome 7 Partial Deletion of Short  
 15 Arm (7p2-), Chromosome 7 Terminal 7p Deletion [del (7) (p21-p22)], Chromosome 8  
 Monosomy 8p2, Chromosome 8 Monosomy 8p21-pter, Chromosome 8 Partial Deletion  
 (short arm), Chromosome 8 Partial Monosomy 8p2, Chromosome 9 Complete Trisomy 9P,  
 Chromosome 9 Partial Deletion of Short Arm, Chromosome 9 Partial Monosomy 9p,  
 Chromosome 9 Partial Monosomy 9p22, Chromosome 9 Partial Monosomy 9p22-pter,  
 20 Chromosome 9 Partial Trisomy 9P Included, Chromosome 9 Ring, Chromosome 9  
 Tetrasomy 9p, Chromosome 9 Tetrasomy 9p Mosaicism, Chromosome 9 Trisomy 9p  
 (Multiple Variants), Chromosome 9 Trisomy 9 (pter-p21 to q32) Included, Chromosome 9  
 Trisomy Mosaic, Chromosome 9 Trisomy Mosaic, Chromosome 10 Distal Trisomy 10q,  
 Chromosome 10 Monosomy, Chromosome 10 Monosomy 10p, Chromosome 10, Partial  
 25 Deletion (short arm), Chromosome 10, 10p- Partial, Chromosome 10 Partial Trisomy  
 10q24-qter, Chromosome 10 Trisomy 10q2, Partial Monosomy of Long Arm of  
 Chromosome 11, Chromosome 11 Partial Monosomy 11q, Chromosome 11 Partial  
 Trisomy, Chromosome 11 Partial Trisomy 11q13-qter, Chromosome 11 Partial Trisomy  
 11q21-qter, Chromosome 11 Partial Trisomy 11q23-qter, Chromosome 11q, Partial  
 30 Trisomy, Chromosome 12 Isochromosome 12p Mosaic, Chromosome 13 Partial  
 Monosomy 13q, Chromosome 13, Partial Monosomy of the Long Arm, Chromosome 14

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Ring, Chromosome 14 Trisomy, Chromosome 15 Distal Trisomy 15q, Chromosome r15,  
 Chromosome 15 Ring, Chromosome 15 Trisomy 15q2, Chromosome 15q, Partial  
 Duplication Syndrome, Chromosome 17 Interstitial Deletion 17p, Chromosome 18 Long  
 Arm Deletion Syndrome, Chromosome 18 Monosomy 18p, Chromosome 18 Monosomy  
 5 18Q, Chromosome 18 Ring, Chromosome 18 Tetrasomy 18p, Chromosome 18q-  
 Syndrome, Chromosome 21 Mosaic 21 Syndrome, Chromosome 21 Ring, Chromosome 21  
 Translocation 21 Syndrome, Chromosome 22 Inverted Duplication (22pter-22q11),  
 Chromosome 22 Partial Trisomy (22pter-22q11), Chromosome 22 Ring, Chromosome 22  
 Trisomy Mosaic, Chromosome 48 XXYY, Chromosome 48 XXXY, Chromosome r15,  
 10 Chromosomal Triplication, Chromosome Triplication, Chromosome Triploidy Syndrome,  
 Chromosome X, Chromosome XXY, Chronic Acholuric Jaundice, Chronic Adhesive  
 Arachnoiditis, Chronic Adrenocortical Insufficiency, Chronic Cavemosis, Chronic  
 Congenital Aregenerative Anemia, Chronic Dysphagocytosis, Chronic Familial  
 Granulomatosis, Chronic Familial Icterus, Chronic Fatigue Immune Dysfunction  
 15 Syndrome (CFIDS), Chronic Granulomatous Disease, Chronic Guillain-Barre Syndrome,  
 Chronic Idiopathic Jaundice, Chronic Idiopathic Polyneuritis (CIP), Chronic Inflammatory  
 Demyelinating Polyneuropathy, Chronic Inflammatory Demyelinating  
 Polyradiculoneuropathy, Chronic Motor Tic, Chronic Mucocutaneous Candidiasis,  
 Chronic Multiple Tics, Chronic Non-Specific Ulcerative Colitis, Chronic Obliterative  
 20 Cholangitis, Chronic Peptic Ulcer and Esophagitis Syndrome, Chronic Progressive Chorea,  
 Chronic Progressive External Ophthalmoplegia Syndrome, Chronic Progressive External  
 Ophthalmoplegia and myopathy, Chronic Progressive External Ophthalmoplegia with  
 Ragged Red Fibers, Chronic Relapsing Polyneuropathy, Chronic Sarcoidosis, Chronic  
 Spasmodic Dysphonia, Chronic Vomiting in Childhood, CHS, Churg-Strauss Syndrome,  
 25 Cicatricial Pemphigoid, CIP, Cirrhosis Congenital Pigmentary, Cirrhosis, Cistinuria,  
 Citrullinemia, CJD, Classic Schindler Disease, Classic Type Pfeiffer Syndrome, Classical  
 Maple Syrup Urine Disease, Classical Hemophilia, Classical Form Cockayne Syndrome  
 Type I (Type A), Classical Leigh's Disease, Classical Phenylketonuria, Classical X-Linked  
 Pelizaeus-Merzbacher Brain Sclerosis, CLE, Cleft Lip/Palate Mucous Cysts Lower Lip PP  
 30 Digital and Genital Anomalies, Cleft Lip-Palate Blepharophimosis Lagophthalmos and  
 Hypertelorism, Cleft Lip/Palate with Abnormal Thumbs and Microcephaly, Cleft palate-

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joint contractures-dandy walker malformations, Cleft Palate and Cleft Lip, Cleidocranial Dysplasia w/ Micrognathia, Absent Thumbs, & Distal Aphyalangia, Cleidocranial Dysostosis, Cleidocranial Dysplasia, Click murmur syndrome, CLN1, Clonic Spasmodic, Clouston Syndrome, Clubfoot, CMDI, CMM, CMT, CMTC, CMTX, COA Syndrome, 5 Coarctation of the aorta, Coats' Disease, Cobblestone dysplasia, Cochin Jewish Disorder, Cockayne Syndrome, COD-MD Syndrome, COD, Coffin Lowry Syndrome, Coffin Syndrome, Coffin Siris Syndrome, COFS Syndrome, Cogan Corneal Dystrophy, Cogan Reese Syndrome, Cohen Syndrome, Cold Agglutinin Disease, Cold Antibody Disease, Cold Antibody Hemolytic Anemia, Colitis Ulcerative, Colitis Gravis, Colitis Ulcerative 10 Chronic Non-Specific Ulcerative Colitis, Collodion Baby, Coloboma Heart Defects Atresia of the Choanae Retardation of Growth and Development Genital and Urinary Anomalies and Ear Anomalies, Coloboma, Colonic Neurosis, Color blindness, Colour blindness, Colpocephaly, Columnar-Like Esophagus, Combined Cone-Rod Degeneration, Combined Immunodeficiency with Immunoglobulins, Combined Mesoectodermal Dysplasia, 15 Common Variable Hypogammaglobulinemia, Common Variable Immunodeficiency, Common Ventricle, Communicating Hydrocephalus, Complete Absence of Hypoxanthine-Guanine Phosphoribosyltransferase, Complete Atrioventricular Septal Defect, Complement Component 1 Inhibitor Deficiency, Complement Component C1 Regulatory Component Deficiency, Complete Heart Block, Complex Carbohydrate Intolerance, Complex Regional 20 Pain Syndrome, Complex V ATP Synthase Deficiency, Complex I, Complex I NADH dehydrogenase deficiency, Complex II, Complex II Succinate dehydrogenase deficiency, Complex III, Complex III Ubiquinone-cytochrome c oxidoreductase deficiency, Complex IV, Complex IV Cytochrome c oxidase deficiency, Complex IV Deficiency, Complex V, Cone-Rod Degeneration, Cone-Rod Degeneration Progressive, Cone Dystrophy, Cone- 25 Rod Dystrophy, Confluent Reticular Papillomatosis, Congenital with low PK Kinetics, Congenital Absence of Abdominal Muscles, Congenital Absence of the Thymus and Parathyroids, Congenital Achromia, Congenital Addison's Disease, Congenital Adrenal Hyperplasia, Congenital Adrenal Hyperplasia, Congenital Afibrinogenemia, Congenital Alveolar Hypoventilation, Congenital Anemia of Newborn, Congenital Bilateral 30 Persylvian Syndrome, Congenital Brown Syndrome, Congenital Cardiovascular Defects, Congenital Central Hypoventilation Syndrome, Congenital Cerebral Palsy, Congenital

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Cervical Synostosis, Congenital Clapsed Thumb with Mental Retardation, Congenital Contractural Arachnodactyly, Congenital Contractures Multiple with Arachnodactyly, Congenital Cyanosis, Congenital Defect of the Skull and Scalp, Congenital Dilatation of Intrahepatic Bile Duct, Congenital Dysmyelinating Neuropathy, Congenital

5 Dysphagocytosis, Congenital Dysplastic Angiectasia, Congenital Erythropoietic Porphyrin, Congenital Factor XIII Deficiency, Congenital Failure of Autonomic Control of Respiration, Congenital Familial Nonhemolytic Jaundice Type I, Congenital Familial Protracted Diarrhea, Congenital Form Cockayne Syndrome Type II (Type B), Congenital Generalized Fibromatosis, Congenital German Measles, Congenital Giant Axonal

10 Neuropathy, Congenital Heart Block, Congenital Heart Defects, Congenital Hemidysplasia with Ichthyosis Erythroderma and Limb Defects, Congenital Hemolytic Jaundice, Congenital Hemolytic Anemia, Congenital Hepatic Fibrosis, Congenital Hereditary Comeal Dystrophy, Congenital Hereditary Lymphedema, Congenital Hyperchondroplasia, Congenital Hypomyelinating Polyneuropathy, Congenital Hypomyelination Neuropathy,

15 Congenital Hypomyelination, Congenital Hypomyelination (Onion Bulb) Polyneuropathy, Congenital Ichthyosiform Erythroderma, Congenital Keratoconus, Congenital Lactic Acidosis, Congenital Lactose Intolerance, Congenital Lipodystrophy, Congenital Liver Cirrhosis, Congenital Lobar Emphysema, Congenital Localized Emphysema, Congenital Macroglossia, Congenital Medullary Stenosis, Congenital Megacolon, Congenital

20 Melanocytic Nevus, Congenital Mesodermal Dysmorphodystrophy, Congenital Mesodermal Dystrophy, Congenital Microvillus Atrophy, Congenital Multiple Arthrogryposis, Congenital Myotonic Dystrophy, Congenital Neuropathy caused by Hypomyelination, Congenital Pancytopenia, Congenital Pernicious Anemia, Congenital Pernicious Anemia due to Defect of Intrinsic Factor, Congenital Pernicious Anemia due to

25 Defect of Intrinsic Factor, Congenital Pigmentary Cirrhosis, Congenital Porphyrin, Congenital Proximal myopathy Associated with Desmin Storage myopathy, Congenital Pulmonary Emphysema, Congenital Pure Red Cell Anemia, Congenital Pure Red Cell Aplasia, Congenital Retinal Blindness, Congenital Retinal Cyst, Congenital Retinitis Pigmentosa, Congenital Retinoschisis, Congenital Rod Disease, Congenital Rubella

30 Syndrome, Congenital Scalp Defects with Distal Limb Reduction Anomalies, Congenital Sensory Neuropathy, Congenital SMA with arthrogryposis, Congenital Spherocytic

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Anemia, Congenital Spondyloepiphyseal Dysplasia, Congenital Tethered Cervical Spinal Cord Syndrome, Congenital Tyrosinosis, Congenital Varicella Syndrome, Congenital Vascular Cavemous Malformations, Congenital Vascular Veils in the Retina, Congenital Word Blindness, Congenital Wandering Spleen (Pediatric), Congestive Cardio myopathy,

5 Conical Cornea, Conjugated Hyperbilirubinemia, Conjunctivitis, Conjunctivitis Ligneous, Conjunctivo-Urethro-Synovial Syndrome, Conn's Syndrome, Connective Tissue Disease, Conradi Disease, Conradi Hunermann Syndrome, Constitutional Aplastic Anemia, Constitutional Erythroid Hypoplasia, Constitutional Eczema, Constitutional Liver Dysfunction, Constitutional Thrombopathy, Constricting Bands Congenital, Constrictive

10 Pericarditis with Dwarfism, Continuous Muscle Fiber Activity Syndrome, Contractural Arachnodactyly, Contractures of Feet Muscle Atrophy and Oculomotor Apraxia, Convulsions, Cooley's anemia, Copper Transport Disease, Coproporphyrria Porphyrria Hepatica, Cor Triatriatum, Cor Triatriatum Sinistrum, Cor Triloculare Batriatum, Cor Biloculare, Cori Disease, Cornea Dystrophy, Corneal Amyloidosis, Corneal Clouding-

15 Cutis Laxa-Mental Retardation, Corneal Dystrophy, Cornelia de Lange Syndrome, Coronal Dentine Dysplasia, Coronary Artery Disease, Coronary Heart Disease, Corpus Callosum Agenesis, Cortical-Basal Ganglionic Degeneration, Corticalis Deformaris, Cortico-Basal Ganglionic Degeneration (CBGD), Corticobasal Degeneration, Corticosterone Methloxidase Deficiency Type I, Corticosterone Methyloxidase Deficiency Type II,

20 Cortisol, Costello Syndrome, Cot Death, COVESDEM Syndrome, COX, COX Deficiency, COX Deficiency French-Canadian Type, COX Deficiency Infantile Mitochondrial myopathy de Toni-Fanconi-Debre included, COX Deficiency Type Benign Infantile Mitochondrial Myopathy, CP, CPEO, CPEO with myopathy, CPEO with Ragged-Red Fibers, CPPD Familial Form, CPT Deficiency, CPTD, Cranial Arteritis, Cranial

25 Meningoencephalocele, Cranio-Oro-Digital Syndrome, Craniocarpotarsal dystrophy, Craniocoele, Craniodigital Syndrome-Mental Retardation Scott Type, Craniofacial Dysostosis, Craniofacial Dysostosis-PD Arteriosus-Hypertrichosis-Hypoplasia of Labia, Craniofrontonasal Dysplasia, Craniometaphyseal Dysplasia, Cranioorodigital Syndrome, Cranioorodigital Syndrome Type II, Craniostenosis Crouzon Type, Craniostenosis,

30 Craniosynostosis-Choanal Atresia-Radial Humeral Synostosis, Craniosynostosis-Hypertrichosis-Facial and Other Anomalies, Craniosynostosis Midfacial Hypoplasia and

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Foot Abnormalities, Craniosynostosis Primary, Craniosynostosis-Radial Aplasia Syndrome, Craniosynostosis with Radial Defects, Cranium Bifidum, CREST Syndrome, Creutzfeldt Jakob Disease, Cri du Chat Syndrome, Crib Death, Crigler Najjar Syndrome Type I, Crohn's Disease, Cronkhite-Canada Syndrome, Cross Syndrome, Cross' Syndrome, Cross-McKusick-Breen Syndrome, Crouzon, Crouzon Syndrome, Crouzon Craniofacial Dysostosis, Cryoglobulinemia Essential Mixed, Cryptophthalmos-Syndactyly Syndrome, Cryptorchidism-Dwarfism-Subnormal Mentality, Crystalline Corneal Dystrophy of Schnyder, CS, CSD, CSID, CSO, CST Syndrome, Curly Hair-Ankyloblepharon-Nail Dysplasia, Curschmann-Batten-Steinert Syndrome, Curth Macklin Type Ichthyosis Hystrix, Curth-Macklin Type, Cushing's, Cushing Syndrome, Cushing's III, Cutaneous Malignant Melanoma Hereditary, Cutaneous Porphyrias, Cutis Laxa, Cutis Laxa-Growth Deficiency Syndrome, Cutis Marmorata Telangiectatica Congenita, CVI, CVID, CVS, Cyclic vomiting syndrome, Cystic Disease of the Renal Medulla, Cystic Hygroma, Cystic Fibrosis, Cystic Lymphangioma, Cystine-Lysine-Arginine-Omithinuria, Cystine Storage Disease, Cystinosis, Cystinuria, Cystinuria with Dibasic Aminoaciduria, Cystinuria Type I, Cystinuria Type II, Cystinuria Type III, Cysts of the Renal Medulla Congenital, Cytochrome C Oxidase Deficiency, D.C., Dacryosialoadenopathy, Dacryosialoadenopathia, Dalpro, Dalton, Daltonism, Danbolt-Cross Syndrome, Dancing Eyes-Dancing Feet Syndrome, Dandy-Walker Syndrome, Dandy-Walker Cyst, Dandy-Walker Deformity, Dandy Walker Malformation, Danish Cardiac Type Amyloidosis (Type III), Darier Disease, Davidson's Disease, Davies' Disease, DBA, DBS, DC, DD, De Barsy Syndrome, De Barsy-Moens-Diercks Syndrome, de Lange Syndrome, De Morsier Syndrome, De Santis Cacchione Syndrome, de Toni-Fanconi Syndrome, Deafness Congenital and Functional Heart Disease, Deafness-Dwarfism-Retinal Atrophy, Deafness-Functional Heart Disease, Deafness Onychodystrophy Osteodystrophy and Mental Retardation, Deafness and Pili Torti Bjornstad Type, Deafness Sensorineural with Imperforate Anus and Hypoplastic Thumbs, Debrancher Deficiency, Deciduous Skin, Defect of Enterocyte Intrinsic Factor Receptor, Defect in Natural Killer Lymphocytes, Defect of Renal Reabsorption of Carnitine, Deficiency of Glycoprotein Neuraminidase, Deficiency of Mitochondrial Respiratory Chain Complex IV, Deficiency of Platelet Glycoprotein Ib, Deficiency of Von Willebrand Factor Receptor, Deficiency of Short-

Chain Acyl-CoA Dehydrogenase (ACADS), Deformity with Mesomelic Dwarfism, Degenerative Chorea, Degenerative Lumbar Spinal Stenosis, Degos Disease, Degos-Kohlmeier Disease, Degos Syndrome, DEH, Dejerine-Roussy Syndrome, Dejerine Sottas Disease, Deletion 9p Syndrome Partial, Deletion 11q Syndrome Partial, Deletion 13q Syndrome Partial, Delleman-Oorthuys Syndrome, Delleman Syndrome, Dementia with Lobar Atrophy and Neuronal Cytoplasmic Inclusions, Demyelinating Disease, DeMyer Syndrome, Dentin Dysplasia Coronal, Dentin Dysplasia Radicular, Dentin Dysplasia Type I, Dentin Dysplasia Type II, Dentinogenesis Imperfecta Brandywine type, Dentinogenesis Imperfecta Shields Type, Dentinogenesis Imperfecta Type III, Dento-Oculo- Osseous Dysplasia, Dentooculocutaneous Syndrome, Denys-Drash Syndrome, Depakene, Depakene<sup>TM</sup> exposure, Depakote, Depakote Sprinkle, Depigmentation-Gingival Fibromatosis-Microphthalmia, Dercum Disease, Dermatitis Atopic, Dermatitis Exfoliativa, Dermatitis Herpetiformis, Dermatitis Multiformis, Dermatochalasia Generalized, Dermatolysis Generalized, Dermatomegaly, Dermatomyositis sine myositis, Dermatomyositis, Dermatosparaxis, Dermatostomatitis Stevens Johnson Type, Desbuquois Syndrome, Desmin Storage myopathy, Desquamation of Newborn, Deuteranomaly, Developmental Reading Disorder, Developmental Gerstmann Syndrome, Devergie Disease, Devic Disease, Devic Syndrome, Dextrocardia- Bronchiectasis and Sinusitis, Dextrocardia with Situs Inversus, DGS, DGSX Golabi-Rosen Syndrome Included, DH, DHAP alkyl transferase deficiency, DHBS Deficiency, DHOF, DHPR Deficiency, Diabetes Insipidus, Diabetes Insipidus Diabetes Mellitus Optic Atrophy and Deafness, Diabetes Insipidus Neurohypophyseal, Diabetes Insulin Dependent, Diabetes Mellitus, Diabetes Mellitus Addison's Disease Myxedema, Diabetic Acidosis, Diabetic Bearded Woman Syndrome, Diamond-Blackfan Anemia, Diaphragmatic Apnea, Diaphyseal Aclasis, Diastrophic Dwarfism, Diastrophic Dysplasia, Diastrophic Nanism Syndrome, Dicarboxylic Aminoaciduria, Dicarboxylicaciduria Caused by Defect in Beta-Oxidation of Fatty Acids, Dicarboxylicaciduria due to Defect in Beta-Oxidation of Fatty Acids, Dicarboxylicaciduria due to MCADH Deficiency, Dichromasy, Dicker-Opitz, DIDMOAD, Diencephalic Syndrome, Diencephalic Syndrome of Childhood, Diencephalic Syndrome of Emaciation, Dienoyl-CoA Reductase Deficiency, Diffuse Cerebral Degeneration in Infancy, Diffuse Degenerative Cerebral Disease, Diffuse Idiopathic Skeletal Hyperostosis,

Diffusum-Glycopeptiduria, DiGeorge Syndrome, Digital-Oro-Cranio Syndrome, Digito-Oto-Palatal Syndrome, Digito-Oto-Palatal Syndrome Type I, Digito-Oto-Palatal Syndrome Type II, Dihydrobiopterin Synthetase Deficiency, Dihydropteridine Reductase Deficiency, Dihydroxyacetonephosphate synthase, Dilated (Congestive) Cardio myopathy, Dimitri

5 Disease, Diplegia of Cerebral Palsy, Diplo-Y Syndrome, Disaccharidase Deficiency, Disaccharide Intolerance I, Discoid Lupus, Discoid Lupus Erythematosus, DISH, Disorder of Cornification, Disorder of Cornification Type I, Disorder of Cornification 4, Disorder of Cornification 6, Disorder of Cornification 8, Disorder of Cornification 9 Netherton's Type, Disorder of Cornification 11 Phytanic Acid Type, Disorder of Cornification 12 (Neutral

10 Lipid Storage Type), Disorder of Cornification 13, Disorder of Cornification 14, Disorder of Cornification 14 Trichothiodystrophy Type, Disorder of Cornification 15 (Keratitis Deafness Type), Disorder of Cornification 16, Disorder of Cornification 18 Erythrokeratoderma Variabilis Type, Disorder of Cornification 19, Disorder of Cornification 20, Disorder of Cornification 24, Displaced Spleen, Disseminated Lupus

15 Erythematosus, Disseminated Neurodermatitis, Disseminated Sclerosis, Distal 11q Monosomy, Distal 11q- Syndrome, Distal Arthrogryposis Multiplex Congenita Type IIA, Distal Arthrogryposis Multiplex Congenita Type IIA, Distal Arthrogryposis Type IIA, Distal Arthrogryposis Type 2A, Distal Duplication 6q, Distal Duplication 10q, Dup(10q) Syndrome, Distal Duplication 15q, Distal Monosomy 9p, Distal Trisomy 6q, Distal

20 Trisomy 10q Syndrome, Distal Trisomy 11q, Divalproex, DJS, DKC, DLE, DLPIII, DM, DMC Syndrome, DMC Disease, DMD, DNS Hereditary, DOC I, DOC 2, DOC 4, DOC 6 (Harlequin Type), DOC 8 Curth-Macklin Type, DOC 11 Phytanic Acid Type, DOC 12 (Neutral Lipid Storage Type), DOC 13, DOC 14, DOC 14 Trichothiodystrophy Type, DOC 15 (Keratitis Deafness Type), DOC 16, DOC 16 Unilateral Hemidysplasia Type,

25 DOC 18, DOC 19, DOC 20, DOC 24, Dohle's Bodies-Myelopathy, Dolichospondylic Dysplasia, Dolichostenomelia, Dolichostenomelia Syndrome, Dominant Type Kenny-Caffe Syndrome, Dominant Type Myotonia Congenita, Donahue Syndrome, Donath-Landsteiner Hemolytic Anemia, Donath-Landsteiner Syndrome, DOOR Syndrome, DOORS Syndrome, Dopa-responsive Dystonia (DRD), Dorfman Chanarin Syndrome,

30 Dowling-Meara Syndrome, Down Syndrome, DR Syndrome, Drash Syndrome, DRD, Dreifuss-Emery Type Muscular Dystrophy with Contractures, Dressler Syndrome, Drifting

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Spleen, Drug-induced Acanthosis Nigricans, Drug-induced Lupus Erythematosus, Drug-related Adrenal Insufficiency, Drummond's Syndrome, Dry Beriberi, Dry Eye, DTD, Duane's Retraction Syndrome, Duane Syndrome, Duane Syndrome Type IA 1B and 1C, Duane Syndrome Type 2A 2B and 2C, Duane Syndrome Type 3A 3B and 3C, Dubin

5 Johnson Syndrome, Dubowitz Syndrome, Duchenne, Duchenne Muscular Dystrophy, Duchenne's Paralysis, Duhring's Disease, Duncan Disease, Duncan's Disease, Duodenal Atresia, Duodenal Stenosis, Duodenitis, Duplication 4p Syndrome, Duplication 6q Partial, Dupuy's Syndrome, Dupuytren's Contracture, Dutch-Kennedy Syndrome, Dwarfism, Dwarfism Campomelic, Dwarfism Cortical Thickening of the Tubular Bones & Transient

10 Hypocalcemia, Dwarfism Levi's Type, Dwarfism Metatropic, Dwarfism-Onychodysplasia, Dwarfism-Pericarditis, Dwarfism with Renal Atrophy and Deafness, Dwarfism with Rickets, DWM, Dyggve Melchior Clausen Syndrome, Dysautonomia Familial, Dysbetalipoproteinemia Familial, Dyschondrodysplasia with Hemangiomas, Dyschondrosteosis, Dyschromatosis Universalis Hereditaria, Dysencephalia

15 Splanchnocystica, Dyskeratosis Congenita, Dyskeratosis Congenita Autosomal Recessive, Dyskeratosis Congenita Scoggins Type, Dyskeratosis Congenita Syndrome, Dyskeratosis Follicularis Vegetans, Dyslexia, Dysmyelogenic Leukodystrophy, Dysmyelogenic Leukodystrophy-Megalobare, Dysphonia Spastica, Dysplasia Epiphysialis Punctata, Dysplasia Epiphyseal Hemimelica, Dysplasia of Nails With Hypodontia, Dysplasia

20 Cleidocranial, Dysplasia Fibrous, Dysplasia Gigantism Syndrome X-Linked, Dysplasia Osteodental, Dysplastic Nevus Syndrome, Dysplastic Nevus Type, Dyssynergia Cerebellaris Myoclonica, Dyssynergia Esophagus, Dystonia, Dystopia Canthorum, Dystrophia Adiposogenitalis, Dystrophia Endothelialis Cornea, Dystrophia Mesodermalis, Dystrophic Epidermolysis Bullosa, Dystrophy, Asphyxiating Thoracic, Dystrophy

25 Myotonic, E-D Syndrome, Eagle-Barrett Syndrome, Eales Retinopathy, Eales Disease, Ear Anomalies-Contractures-Dysplasia of Bone with Kyphoscoliosis, Ear Patella Short Stature Syndrome, Early Constraint Defects, Early Hypercalcemia Syndrome with Elfin Facie, Early-onset Dystonia, Eaton Lambert Syndrome, EB, Ebstein's anomaly, EBV Susceptibility (EBVS), EBVS, ECD, ECPSG, Ectodermal Dysplasias, Ectodermal

30 Dysplasia Anhidrotic with Cleft Lip and Cleft Palate, Ectodermal Dysplasia-Exocrine Pancreatic Insufficiency, Ectodermal Dysplasia Rapp-Hodgkin type, Ectodermal and

Mesodermal Dysplasia Congenital, Ectodermal and Mesodermal Dysplasia with Osseous  
 Involvement, Ectodermosis Erosiva Pluriorificialis, Ectopia Lentis, Ectopia Vesicae,  
 Ectopic ACTH Syndrome, Ectopic Adrenocorticotrophic Hormone Syndrome, Ectopic  
 Anus, Ectrodactilia of the Hand, Ectrodactyly, Ectrodactyly-Ectodermal Dysplasia-  
 5 Clefting Syndrome, Ectrodactyly Ectodermal Dysplasias Clefting Syndrome, Ectrodactyly  
 Ectodermal Dysplasia Cleft Lip/Cleft Palate, Eczema, Eczema-Thrombocytopenia-  
 Immunodeficiency Syndrome, EDA, EDMD, EDS, EDS Arterial-Ecchymotic Type, EDS  
 Arthrochalasia, EDS Classic Severe Form, EDS Dysfibronectinemic, EDS Gravis Type,  
 EDS Hypermobility, EDS Kyphoscoliotic, EDS Kyphoscoliosis, EDS Mitis Type, EDS  
 10 Ocular-Scoliotic, EDS Progeroid, EDS Periodontosis, EDS Vascular, EEC Syndrome,  
 EFE, EHBA, EHK, Ehlers Danlos Syndrome, Ehlers-Danlos syndrome, Ehlers Danlos IX,  
 Eisenmenger Complex, Eisenmenger's complex, Eisenmenger Disease, Eisenmenger  
 Reaction, Eisenmenger Syndrome, Ekblom Syndrome, Ekman-Lobstein Disease,  
 Ektrodactyly of the Hand, EKV, Elastin fiber disorders, Elastorrhexis Generalized,  
 15 Elastosis Dystrophica Syndrome, Elective Mutism (obsolete), Elective Mutism,  
 Electrocardiogram (ECG or EKG), Electron Transfer Flavoprotein (ETF) Dehydrogenase  
 Deficiency: (GAIL & MADD), Electrophysiologic study (EPS), Elephant Nails From Birth,  
 Elephantiasis Congenita Angiomatosa, Hemangiectatic Hypertrophy, Elfin Facies with  
 Hypercalcemia, Ellis-van Creveld Syndrome, Ellis Van Creveld Syndrome, Embryoma  
 20 Kidney, Embryonal Adenomyosarcoma Kidney, Embryonal Carcinosarcoma Kidney,  
 Embryonal Mixed Tumor Kidney, EMC, Emery Dreyfus Muscular Dystrophy, Emery-  
 Dreifuss Muscular Dystrophy, Emery-Dreifuss Syndrome, EMF, EMG Syndrome, Empty  
 Sella Syndrome, Encephalitis Periaxialis Diffusa, Encephalitis Periaxialis Concentrica,  
 Encephalocele, Encephalofacial Angiomatosis, Encephalopathy, Encephalotrigeminal  
 25 Angiomatosis, Enchondromatosis with Multiple Cavemous Hemangiomas, Endemic  
 Polyneuritis, Endocardial Cushion Defect, Endocardial Cushion Defects, Endocardial  
 Dysplasia, Endocardial Fibroelastosis (EFE), Endogenous Hypertriglyceridemia,  
 Endolymphatic Hydrops, Endometrial Growths, Endometriosis, Endomyocardial Fibrosis,  
 Endothelial Corneal Dystrophy Congenital, Endothelial Epithelial Corneal Dystrophy,  
 30 Endothelium, Engelmann Disease, Enlarged Tongue, Enterocolitis, Enterocyte Cobalamin  
 Malabsorption, Eosinophilia Syndrome, Eosinophilic Cellulitis, Eosinophilic Fasciitis,

Eosinophilic Granuloma, Eosinophilic Syndrome, Epidermal Nevus Syndrome,  
 Epidermolysis Bullosa, Epidermolysis Bullosa Acquisita, Epidermolysis Bullosa  
 Hereditaria, Epidermolysis Bullosa Letalias, Epidermolysis Hereditaria Tarda,  
 Epidermolytic Hyperkeratosis, Epidermolytic Hyperkeratosis (Bullous CIE), Epilepsia  
 5 Procursiva, Epilepsy, Epinephrine, Epiphyseal Changes and High Myopia, Epiphyseal  
 Osteochondroma Benign, Epiphysealis Hemimelica Dysplasia, Episodic-Abnormal Eye  
 Movement, Epithelial Basement Membrane Corneal Dystrophy, Epithelial Corneal  
 Dystrophy of Meesmann Juvenile, Epitheliomatosis Multiplex with Nevus, Epithelium,  
 Epival, EPS, Epstein-Barr Virus-Induced Lymphoproliferative Disease in Males, Erb-  
 10 Goldflam syndrome, Erdheim Chester Disease, Erythema Multiforme Exudativum,  
 Erythema Polymorphe Stevens Johnson Type, Erythroblastophthisis, Erythroblastosis  
 Fetalis, Erythroblastosis Neonatorum, Erythroblastotic Anemia of Childhood, Erythrocyte  
 Phosphoglycerate Kinase Deficiency, Erythrocytosis Imperfecta, Erythrokeratodermia  
 Progressiva Symmetrica, Erythrokeratodermia Progressiva Symmetrica Ichthyosis,  
 15 Erythrokeratodermia Variabilis, Erythrokeratodermia Variabilis Type, Erythrokeratolysis  
 Hiemalis, Erythropoietic Porphyrias, Erythropoietic Porphyria, Escobar Syndrome,  
 Esophageal Atresia, Esophageal Aperistalsis, Esophagitis-Peptic Ulcer, Esophagus Atresia  
 and/or Tracheoesophageal Fistula, Essential Familial Hyperlipemia, Essential Fructosuria,  
 Essential Hematuria, Essential Hemorrhagic Thrombocythemia, Essential Mixed  
 20 Cryoglobulinemia, Essential Moschowitz Disease, Essential Thrombocythemia, Essential  
 Thrombocytopenia, Essential Thrombocytosis, Essential Tremor, Esterase Inhibitor  
 Deficiency, Estren-Dameshek variant of Fanconi Anemia, Estrogen-related Cholestasis,  
 ET, ETF, Ethylmalonic Adipicaciduria, Eulenburg Disease, pc, EVCS, Exaggerated Startle  
 Reaction, Exencephaly, Exogenous Hypertriglyceridemia, Exomphalos-Macroglossia-  
 25 Gigantism Syndrom, Exophthalmic Goiter, Expanded Rubella Syndrome, Exstrophy of the  
 Bladder, EXT, External Chondromatosis Syndrome, Extrahepatic Biliary Atresia,  
 Extramedullary Plasmacytoma, Exudative Retinitis, Eye Retraction Syndrome, FA1, FAA,  
 Fabry Disease, FAC, FACB, FACD, FACE, FACF, FACG, FACH, Facial Nerve Palsy,  
 Facial Paralysis, Facial Ectodermal Dysplasias, Facial Ectodermal Dysplasia, Facio-  
 30 Scapulo-Humeral Dystrophy, Facio-Auriculo-Vertebral Spectrum, Facio-cardio-cutaneous  
 syndrome, Facio-Fronto-Nasal Dysplasia, Faciocutaneoskeletal Syndrome,

Faciodigitogenital syndrome, Faciogenital dysplasia, Faciogenitopopliteal Syndrome,  
 Faciopalatoosseous Syndrome, Faciopalatoosseous Syndrome Type II,  
 Facioscapulohumeral muscular dystrophy, Factitious Hypoglycemia, Factor VIII  
 Deficiency, Factor IX Deficiency, Factor XI Deficiency, Factor XII deficiency, Factor XIII  
 5 Deficiency, Fahr Disease, Fahr's Disease, Failure of Secretion Gastric Intrinsic Factor,  
 Fairbank Disease, Fallot's Tetralogy, Familial Acrogeria, Familial Acromicria, Familial  
 Adenomatous Colon Polyposis, Familial Adenomatous Polyposis with Extraintestinal  
 Manifestations, Familial Alobar Holoprosencephaly, Familial Alpha-Lipoprotein  
 Deficiency, Familial Amyotrophic Chorea with Acanthocytosis, Familial Arrhythmic  
 10 Myoclonus, Familial Articular Chondrocalcinosis, Familial Atypical Mole-Malignant  
 Melanoma Syndrome, Familial Broad Beta Disease, Familial Calcium Gout, Familial  
 Calcium Pyrophosphate Arthropathy, Familial Chronic Obstructive Lung Disease, Familial  
 Continuous Skin Peeling, Familial Cutaneous Amyloidosis, Familial Dysproteinemia,  
 Familial Emphysema, Familial Enteropathy Microvillus, Familial Foveal Retinoschisis,  
 15 Familial Hibernation Syndrome, Familial High Cholesterol, Familial Hemochromatosis,  
 Familial High Blood Cholesterol, Familial High-Density Lipoprotein Deficiency, Familial  
 High Serum Cholesterol, Familial Hyperlipidemia, Familial Hypoproteinemia with  
 Lymphangietatic Enteropathy, Familial Jaundice, Familial Juvenile Nephronophthisis-  
 Associated Ocular Anomaly, Familial Lichen Amyloidosis (Type IX), Familial Lumbar  
 20 Stenosis, Familial Lymphedema Praecox, Familial Mediterranean Fever, Familial Multiple  
 Polyposis, Familial Nuchal Bleb, Familial Paroxysmal Polyserositis, Familial Polyposis  
 Coli, Familial Primary Pulmonary Hypertension, Familial Renal Glycosuria, Familial  
 Splenic Anemia, Familial Startle Disease, Familial Visceral Amyloidosis (Type VIII),  
 FAMMM, FANCA, FANCB, FANCC, FANCD, FANCE, Fanconi Panmyelopathy,  
 25 Fanconi Pancytopenia, Fanconi II, Fanconi's Anemia, Fanconi's Anemia Type I, Fanconi's  
 Anemia Complementation Group, Fanconi's Anemia Complementation Group A,  
 Fanconi's Anemia Complementation Group B, Fanconi's Anemia Complementation Group  
 C, Fanconi's Anemia Complementation Group D, Fanconi's Anemia Complementation  
 Group E, Fanconi's Anemia Complementation Group G, Fanconi's Anemia  
 30 Complementation Group H, Fanconi's Anemia Estren-Dameshek Variant, FANF, FANG,  
 FANH, FAP, FAPG, Farber's Disease, Farber's Lipogranulomatosis, FAS, Fasting

Hypoglycemia, Fat-Induced Hyperlipemia, Fatal Granulomatous Disease of Childhood, Fatty Oxidation Disorders, Fatty Liver with Encephalopathy, FAV, FCH, FCMD, FCS Syndrome, FD, FDH, Febrile Mucocutaneous Syndrome Stevens Johnson Type, Febrile Neutrophilic Dermatositis Acute, Febrile Seizures, Feinberg's syndrome, Feissinger-Leroy-Reiter Syndrome, Female Pseudo-Turner Syndrome, Femoral Dysgenesis Bilateral-Robin Anomaly, Femoral Dysgenesis Bilateral, Femoral Facial Syndrome, Femoral Hypoplasia-Unusual Facies Syndrome, Fetal Alcohol Syndrome, Fetal Anti-Convulsant Syndrome, Fetal Cystic Hygroma, Fetal Effects of Alcohol, Fetal Effects of Chickenpox, Fetal Effects of Thalidomide, Fetal Effects of Varicella Zoster Virus, Fetal Endomyocardial Fibrosis, Fetal Face Syndrome, Fetal Iritis Syndrome, Fetal Transfusion Syndrome, Fetal Valproate Syndrome, Fetal Valproic Acid Exposure Syndrome, Fetal Varicella Infection, Fetal Varicella Zoster Syndrome, FFDD Type II, FG Syndrome, FGDY, FHS, Fibrin Stabilizing Factor Deficiency, Fibrinase Deficiency, Fibrinoid Degeneration of Astrocytes, Fibrinoid Leukodystrophy, Fibrinoligase Deficiency, Fibroblastoma Perineural, Fibrocystic Disease of Pancreas, Fibrodysplasia Ossificans Progressiva, Fibroelastic Endocarditis, Fibromyalgia, Fibromyalgia-Fibromyositis, Fibromyositis, Fibrosing Cholangitis, Fibrositis, Fibrous Ankylosis of Multiple Joints, Fibrous Cavemosis, Fibrous Dysplasia, Fibrous Plaques of the Penis, Fibrous Sclerosis of the Penis, Fickler-Winkler Type, Fiedler Disease, Fifth Digit Syndrome, Filippi Syndrome, Finnish Type Amyloidosis (Type V), First Degree Congenital Heart Block, First and Second Branchial Arch Syndrome, Fischer's Syndrome, Fish Odor Syndrome, Fissured Tongue, Flat Adenoma Syndrome, Flatau-Schilder Disease, Flavin Containing Monooxygenase 2, Floating Beta Disease, Floating-Harbor Syndrome, Floating Spleen, Floppy Infant Syndrome, Floppy Valve Syndrome, Fluent aphasia, FMD, FMF, FMO Adult Liver Form, FMO2, FND, Focal Dermal Dysplasia Syndrome, Focal Dermal Hypoplasia, Focal Dermato-Phalangeal Dysplasia, Focal Dystonia, Focal Epilepsy, Focal Facial Dermal Dysplasia Type II, Focal Neuromyotonia, FODH, Folling Syndrome, Fong Disease, FOP, Forbes Disease, Forbes-Albright Syndrome, Forestier's Disease, Forsius-Eriksson Syndrome (X-Linked), Fothergill Disease, Fountain Syndrome, Foveal Dystrophy Progressive, FPO Syndrome Type II, FPO, Fraccaro Type Achondrogenesis (Type IB), Fragile X syndrome, Franceschetti-Zwahlen-Klein Syndrome, Francois Dyscephaly Syndrome, Francois-Neetens

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Speckled Dystrophy, Flecked Corneal Dystrophy, Fraser Syndrome, FRAXA, FRDA, Fredrickson Type I Hyperlipoproteinemia, Freeman-Sheldon Syndrome, Freire-Maia Syndrome, Frey's Syndrome, Friedreich's Ataxia, Friedreich's Disease, Friedreich's Tabes, FRNS, Froelich's Syndrome, Frommel-Chiari Syndrome, Frommel-Chiari  
 5 Syndrome Lactation-Uterus Atrophy, Frontodigital Syndrome, Frontofacionasal Dysostosis, Frontofacionasal Dysplasia, Frontonasal Dysplasia, Frontonasal Dysplasia with Coronal Craniosynostosis, Fructose-1-Phosphate Aldolase Deficiency, Fructosemia, Fructosuria, Fryns Syndrome, FSH, FSHD, FSS, Fuchs Dystrophy, Fucosidosis Type 1, Fucosidosis Type 2, Fucosidosis Type 3, Fukuhara Syndrome, Fukuyama Disease,  
 10 Fukuyama Type Muscular Dystrophy, Fumarylacetoacetase deficiency, Furrowed Tongue, G Syndrome, G6PD Deficiency, G6PD, GA I, GA IIB, GA IIA, GA II, GAI & MADD, Galactorrhea-Amenorrhea Syndrome Nonpuerperal, Galactorrhea-Amenorrhea without Pregnancy, Galactosamine-6-Sulfatase Deficiency, Galactose-1-Phosphate Uridyl Transferase Deficiency, Galactosemia, GALB Deficiency, Galloway-Mowat Syndrome,  
 15 Galloway Syndrome, GALT Deficiency, Gammaglobulin Deficiency, GAN, Ganglioside Neuraminidase Deficiency, Ganglioside Sialidase Deficiency, Gangliosidosis GM1 Type 1, Gangliosidosis GM2 Type 2, Gangliosidosis Beta Hexosaminidase B Deficiency, Gardner Syndrome, Gargoylism, Garies-Mason Syndrome, Gasser Syndrome, Gastric Intrinsic Factor Failure of Secretion, Enterocyte Cobalamin, Gastrinoma, Gastritis,  
 20 Gastroesophageal Laceration-Hemorrhage, Gastrointestinal Polyposis and Ectodermal Changes, Gastroschisis, Gaucher Disease, Gaucher-Schlagenhauser, Gayet-Wernicke Syndrome, GBS, GCA, GCM Syndrome, GCPS, Gee-Herter Disease, Gee-Thaysen Disease, Gehrig's Disease, Gelineau's Syndrome, Gence-Wiedemann Syndrome, Generalized Dystonia, Generalized Familial Neuromyotonia, Generalized Fibromatosis,  
 25 Generalized Flexion Epilepsy, Generalized Glycogenosis, Generalized Hyperhidrosis, Generalized Lipofuscinosis, Generalized Myasthenia Gravis, Generalized Myotonia, Generalized Sporadic Neuromyotonia, Genetic Disorders, Genital Defects, Genital and Urinary Tract Defects, Gerstmann Syndrome, Gerstmann Tetrad, GHBP, GHD, GHR, Giant Axonal Disease, Giant Axonal Neuropathy, Giant Benign Lymphoma, Giant Cell  
 30 Glioblastoma Astrocytoma, Giant Cell Arteritis, Giant Cell Disease of the Liver, Giant Cell Hepatitis, Giant Cell of Newborns Cirrhosis, Giant Cyst of the Retina, Giant Lymph

Node Hyperplasia, Giant Platelet Syndrome Hereditary, Giant Tongue, gic Macular Dystrophy, Gilbert's Disease, Gilbert Syndrome, Gilbert-Dreyfus Syndrome, Gilbert-Lereboullet Syndrome, Gilford Syndrome, Gilles de la Tourette's syndrome, Gillespie Syndrome, Gingival Fibromatosis-Abnormal Fingers Nails Nose Ear Splenomegaly, GLA

5 Deficiency, GLA, GLB1, Glioma Retina, Global aphasia, Globoid Leukodystrophy, Glossoptosis Micrognathia and Cleft Palate, Glucocerebrosidase deficiency, Glucocerebrosidosis, Glucose-6-Phosphate Dehydrogenase Deficiency, Glucose-6-Phosphate Transport Defect, Glucose-6-Phosphate Translocase Deficiency, Glucose-6-Phosphatase Deficiency, Glucose-Galactose Malabsorption, Glucosyl Ceramide Lipidosis,

10 Glutaric Aciduria I, Glutaric Acidemia I, Glutaric Acidemia II, Glutaric Aciduria II, Glutaric Aciduria Type II, Glutaric Aciduria Type III, Glutaricacidemia I, Glutaricacidemia II, Glutaricaciduria I, Glutaricaciduria II, Glutaricaciduria Type IIA, Glutaricaciduria Type IIB, Glutaryl-CoA Dehydrogenase Deficiency, Glutaurate-Aspartate Transport Defect, Gluten-Sensitive Enteropathy, Glycogen Disease of Muscle Type VII,

15 Glycogen Storage Disease I, Glycogen Storage Disease III, Glycogen Storage Disease IV, Glycogen Storage Disease Type V, Glycogen Storage Disease VI, Glycogen Storage Disease VII, Glycogen Storage Disease VIII, Glycogen Storage Disease Type II, Glycogen Storage Disease-Type II, Glycogenosis, Glycogenosis Type I, Glycogenosis Type IA, Glycogenosis Type IB, Glycogenosis Type II, Glycogenosis Type II, Glycogenosis Type

20 III, Glycogenosis Type IV, Glycogenosis Type V, Glycogenosis Type VI, Glycogenosis Type VII, Glycogenosis Type VIII, Glycolic Aciduria, Glycolipid Lipidosis, GM2 Gangliosidosis Type 1, GM2 Gangliosidosis Type 1, GNPTA, Goitrous Autoimmune Thyroiditis, Goldenhar Syndrome, Goldenhar-Gorlin Syndrome, Goldscheider's Disease, Goltz Syndrome, Goltz-Gorlin Syndrome, Gonadal Dysgenesis 45 X, Gonadal Dysgenesis

25 XO, Goniodysgenesis-Hypodontia, Goodman Syndrome, Goodman, Goodpasture Syndrome, Gordon Syndrome, Gorlin's Syndrome, Gorlin-Chaudhry-Moss Syndrome, Gottron Erythrokeratoderma Congenitalis Progressiva Symmetrica, Gottron's Syndrome, Gougerot-Carteaud Syndrome, Grand Mal Epilepsy, Granular Type Corneal Dystrophy, Granulomatous Arteritis, Granulomatous Colitis, Granulomatous Dermatitis with

30 Eosinophilia, Granulomatous Ileitis, Graves Disease, Graves' Hyperthyroidism, Graves' Disease, Greig Cephalopolysyndactyly Syndrome, Groenouw Type I Corneal Dystrophy,

- Groenouw Type II Corneal Dystrophy, Gronblad-Strandberg Syndrome, Grotton Syndrome, Growth Hormone Receptor Deficiency, Growth Hormone Binding Protein Deficiency, Growth Hormone Deficiency, Growth-Mental Deficiency Syndrome of Myhre, Growth Retardation-Rieger Anomaly, GRS, Gruber Syndrome, GS, GSD6, GSD8, GTS,
- 5 Guanosine Triphosphate-Cyclohydrolase Deficiency, Guanosine Triphosphate-Cyclohydrolase Deficiency, Guenther Porphyria, Guerin-Stem Syndrome, Guillain-Barré, Guillain-Barre Syndrome, Gunther Disease, H Disease, H. Gottron's Syndrome, Habit Spasms, HAE, Hageman Factor Deficiency, Hageman factor, Haim-Munk Syndrome, Hajdu-Cheney Syndrome, Hajdu Cheney, HAL Deficiency, Hall-Pallister Syndrome,
- 10 Hallermann-Streiff-Francois syndrome, Hallermann-Streiff Syndrome, Hallervorden-Spatz Disease, Hallervorden-Spatz Syndrome, Hallopeau-Siemens Disease, Hallux Duplication Postaxial Polydactyly and Absence of Corpus Callosum, Halushi-Behcet's Syndrome, Hamartoma of the Lymphatics, Hand-Schueller-Christian Syndrome, HANE, Hanhart Syndrome, Happy Puppet Syndrome, Harada Syndrome, HARD +/-E Syndrome, HARD
- 15 Syndrome, Hare Lip, Harlequin Fetus, Harlequin Type DOC 6, Harlequin Type Ichthyosis, Harley Syndrome, Harrington Syndrome, Hart Syndrome, Hartnup Disease, Hartnup Disorder, Hartnup Syndrome, Hashimoto's Disease, Hashimoto-Pritzker Syndrome, Hashimoto's Syndrome, Hashimoto's Thyroiditis, Hashimoto-Pritzker Syndrome, Hay Well's Syndrome, Hay-Wells Syndrome of Ectodermal Dysplasia, HCMM, HCP, HCTD,
- 20 HD, Heart-Hand Syndrome (Holt-Oram Type), Heart Disease, Hecht Syndrome, HED, Heerferdt-Waldenstrom and Lofgren's Syndromes, Hegglin's Disease, Heinrichsbauer Syndrome, Hemangiomas, Hemangioma Familial, Hemangioma-Thrombocytopenia Syndrome, Hemangiomatosis Chondrodystrophica, Hemangiomatous Branchial Clefts-Lip Pseudocleft Syndrome, Hemifacial Microsomia, Hemimegalencephaly, Hemiparesis of
- 25 Cerebral Palsy, Hemiplegia of Cerebral Palsy, Hemisection of the Spinal Cord, Hemochromatosis, Hemochromatosis Syndrome, Hemodialysis-Related Amyloidosis, Hemoglobin Lepore Syndromes, Hemolytic Anemia of Newborn, Hemolytic Cold Antibody Anemia, Hemolytic Disease of Newborn, Hemolytic-Uremic Syndrome, Hemophilia, Hemophilia A, Hemophilia B, Hemophilia B Factor IX, Hemophilia C,
- 30 Hemorrhagic Dystrophic Thrombocytopenia, Hemorrhagica Aleukia, Hemosiderosis, Hepatic Fructokinase Deficiency, Hepatic Phosphorylase Kinase Deficiency, Hepatic

- Porphyrria, Hepatic Porphyrrias, Hepatic Veno-Occlusive Diseases, Hepato-Renal Syndrome,  
 Hepatolenticular Degeneration, Hepatophosphorylase Deficiency, Hepatorenal  
 Glycogenosis, Hepatorenal Syndrome, Hepatorenal Tyrosinemia, Hereditary  
 Acromelalgia, Hereditary Alkaptonuria, Hereditary Amyloidosis, Hereditary Angioedema,  
 5 Hereditary Areflexic Dystasia, Heredopathia Atactica Polyneuritiformis, Hereditary  
 Ataxia, Hereditary Ataxia Friedrich's Type, Hereditary Benign Acanthosis Nigricans,  
 Hereditary Cerebellar Ataxia, Hereditary Chorea, Hereditary Chronic Progressive Chorea,  
 Hereditary Connective Tissue Disorders, Hereditary Coproporphyrria, Hereditary  
 Coproporphyrria Porphyrria, Hereditary Cutaneous Malignant Melanoma, Hereditary  
 10 Deafness-Retinitis Pigmentosa, Heritable Disorder of Zinc Deficiency, Hereditary DNS,  
 Hereditary Dystopic Lipidosis, Hereditary Emphysema, Hereditary Fructose Intolerance,  
 Hereditary Hemorrhagic Telangiectasia, Hereditary Hemorrhagic Telangiectasia Type I,  
 Hereditary Hemorrhagic Telangiectasia Type II, Hereditary Hemorrhagic Telangiectasia  
 Type III, Hereditary Hyperuricemia and Choreoathetosis Syndrome, Hereditary  
 15 Leptocytosis Major, Hereditary Leptocytosis Minor, Hereditary Lymphedema, Hereditary  
 Lymphedema Tarda, Hereditary Lymphedema Type I, Hereditary Lymphedema Type II,  
 Hereditary Motor Sensory Neuropathy, Hereditary Motor Sensory Neuropathy I,  
 Hereditary Motor Sensory Neuropathy Type III, Hereditary Nephritis, Hereditary Nephritis  
 and Nerve Deafness, Hereditary Nephropathic Amyloidosis, Hereditary Nephropathy and  
 20 Deafness, Hereditary Nonpolyposis Colorectal Cancer, Hereditary Nonpolyposis  
 Colorectal Carcinoma, Hereditary Nonspherocytic Hemolytic Anemia, Hereditary  
 Onchoosteodysplasia, Hereditary Optic Neuroretinopathy, Hereditary Polyposis Coli,  
 Hereditary Sensory and Autonomic Neuropathy Type I, Hereditary Sensory and  
 Autonomic Neuropathy Type II, Hereditary Sensory and Autonomic Neuropathy Type III,  
 25 Hereditary Sensory Motor Neuropathy, Hereditary Sensory Neuropathy type I, Hereditary  
 Sensory Neuropathy Type I, Hereditary Sensory Neuropathy Type II, Hereditary Sensory  
 Neuropathy Type III, Hereditary Sensory Radicular Neuropathy Type I, Hereditary  
 Sensory Radicular Neuropathy Type I, Hereditary Sensory Radicular Neuropathy Type II,  
 Hereditary Site Specific Cancer, Hereditary Spherocytic Hemolytic Anemia, Hereditary  
 30 Spherocytosis, Hereditary Tyrosinemia Type I, Heritable Connective Tissue Disorders,  
 Herlitz Syndrome, Hermans-Herzberg Phakomatosis, Hermansky-Pudlak Syndrome,

Hermaphroditism, Herpes Zoster, Herpes Iris Stevens-Johnson Type, Hers Disease,  
 Heterozygous Beta Thalassemia, Hexoaminidase Alpha-Subunit Deficiency (Variant B),  
 Hexoaminidase Alpha-Subunit Deficiency (Variant B), HFA, HFM, HGPS, HH, HHHO,  
 HHRH, HHT, Hiatal Hernia-Microcephaly-Nephrosis Galloway Type, Hidradenitis  
 5 Suppurativa, Hidrosadenitis Axillaris, Hidrosadenitis Suppurativa, Hidrotic Ectodermal  
 Dysplasias, HIE Syndrome, High Imperforate Anus, High Potassium, High Scapula, HIM,  
 Hirschsprung's Disease, Hirschsprung's Disease Acquired, Hirschsprung Disease  
 Polydactyly of Ulnar & Big Toe and VSD, Hirschsprung Disease with Type D  
 Brachydactyly, Hirsutism, HIS Deficiency, Histidine Ammonia-Lyase (HAL) Deficiency,  
 10 Histidase Deficiency, Histidinemia, Histiocytosis, Histiocytosis X, HLHS, HLP Type II,  
 HMG, HMI, HMSN I, HNHA, HOCM, Hodgkin Disease, Hodgkin's Disease, Hodgkin's  
 Lymphoma, Hollaender-Simons Disease, Holmes-Adie Syndrome, Holocarboxylase  
 Synthetase Deficiency, Holoprosencephaly, Holoprosencephaly Malformation Complex,  
 Holoprosencephaly Sequence, Holt-Oram Syndrome, Holt-Oram Type Heart-Hand  
 15 Syndrome, Homocystinemia, Homocystinuria, Homogentisic Acid Oxidase Deficiency,  
 Homogentisic Aciduria, Homozygous Alpha-1-Antitrypsin Deficiency, HOOD, Horner  
 Syndrome, Horton's disease, HOS, HOS1, Houston-Harris Type Achondrogenesis (Type  
 IA), HPS, HRS, HS, HSAN Type I, HSAN Type II, HSAN-III, HSMN, HSMN Type III,  
 HSN I, HSN-III, Huebner-Herter Disease, Hunner's Patch, Hunner's Ulcer, Hunter  
 20 Syndrome, Hunter-Thompson Type Acromesomelic Dysplasia, Huntington's Chorea,  
 Huntington's Disease, Hurler Disease, Hurler Syndrome, Hurler-Scheie Syndrome, HUS,  
 Hutchinson-Gilford Progeria Syndrome, Hutchinson-Gilford Syndrome, Hutchinson-  
 Weber-Peutz Syndrome, Hutterite Syndrome Bowen-Conradi Type, Hyaline  
 Panneuropathy, Hydranencephaly, Hydrocephalus, Hydrocephalus Agyria and Retinal  
 25 Dysplasia, Hydrocephalus Internal Dandy-Walker Type, Hydrocephalus  
 Noncommunicating Dandy-Walker Type, Hydrocephaly, Hydronephrosis With Peculiar  
 Facial Expression, Hydroxylase Deficiency, Hygroma Colli, Hyper-IgE Syndrome, Hyper-  
 IgM Syndrome, Hyperaldosteronism, Hyperaldosteronism With Hypokalemic Alkalosis,  
 Hyperaldosteronism Without Hypertension, Hyperammonemia, Hyperammonemia Due to  
 30 Carbamylphosphate Synthetase Deficiency, Hyperammonemia Due to Ornithine  
 Transcarbamylase Deficiency, Hyperammonemia Type II, Hyper-Beta Carnosinemia,

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Hyperbilirubinemia I, Hyperbilirubinemia II, Hypercalcemia Familial with Nephrocalcinosis and Indicanuria, Hypercalcemia-Supraaortic Aortic Stenosis, Hypercalciuric Rickets, Hypercapnic acidosis, Hypercatabolic Protein-Losing Enteropathy, Hyperchloremic acidosis, Hypercholesterolemia, Hypercholesterolemia Type IV, 5 Hyperchylomicronemia, Hypercystinuria, Hyperekplexia, Hyperextensible joints, Hyperglobulinemic Purpura, Hyperglycinemia with Ketoacidosis and Lactic Acidosis Propionic Type, Hyperglycinemia Nonketotic, Hypergonadotropic Hypogonadism, Hyperimmunoglobulin E Syndrome, Hyperimmunoglobulin E-Recurrent Infection Syndrome, Hyperimmunoglobulinemia E-Staphylococcal, Hyperkalemia, Hyperkinetic 10 Syndrome, Hyperlipemic Retinitis, Hyperlipidemia I, Hyperlipidemia IV, Hyperlipoproteinemia Type I, Hyperlipoproteinemia Type III, Hyperlipoproteinemia Type IV, Hyperoxaluria, Hyperphalangy-Clinodactyly of Index Finger with Pierre Robin Syndrome, Hyperphenylalanemia, Hyperplastic Epidermolysis Bullosa, Hyperpnea, Hyperpotassemia, Hyperprebeta-Lipoproteinemia, Hyperprolinemia Type I, 15 Hyperprolinemia Type II, Hypersplenism, Hypertelorism with Esophageal Abnormalities and Hypospadias, Hypertelorism-Hypospadias Syndrome, Hypertrophic Cardio myopathy, Hypertrophic Interstitial Neuropathy, Hypertrophic Interstitial Neuritis, Hypertrophic Interstitial Radiculoneuropathy, Hypertrophic Neuropathy of Refsum, Hypertrophic Obstructive Cardio myopathy, Hyperuricemia Chorea, Self-mutilation Syndrome, 20 Hyperuricemia-Oligophrenia, Hypervalinemia, Hypocalcified (Hypomineralized) Type, Hypochondrogenesis, Hypochondroplasia, Hypogammaglobulinemia, Hypogammaglobulinemia Transient of Infancy, Hypogenital Dystrophy with Diabetic Tendency, Hypoglossia-Hypodactylia Syndrome, Hypoglycemia, Exogenous Hypoglycemia, Hypoglycemia with Macroglossia, Hypoglycosylation Syndrome Type 1a, 25 Hypoglycosylation Syndrome Type 1a, Hypogonadism with Anosmia, Hypogonadotropic Hypogonadism and Anosmia, Hypohidrotic Ectodermal Dysplasia, Hypohidrotic Ectodermal Dysplasia Autosomal Dominant type, Hypohidrotic Ectodermal Dysplasias Autorecessive, Hypokalemia, Hypokalemic Alkalosis with Hypercalciuria, Hypokalemic Syndrome, Hypolactasia, Hypomaturation Type (Snow-Capped Teeth), Hypomelanosis of 30 Ito, Hypomelia-Hypotrichosis-Facial Hemangioma Syndrome, Hypomyelination Neuropathy, Hypoparathyroidism, Hypophosphatasia, Hypophosphatemic Rickets with

Hypercalcemia, Hypopigmentation, Hypopigmented macular lesion, Hypoplasia of the  
 Depressor Anguli Oris Muscle with Cardiac Defects, Hypoplastic Anemia, Hypoplastic  
 Congenital Anemia, Hypoplastic Chondrodystrophy, Hypoplastic Enamel-Onycholysis-  
 Hypohidrosis, Hypoplastic (Hypoplastic-Explastic) Type, Hypoplastic Left Heart  
 5 Syndrome, Hypoplastic-Triphalangeal Thumbs, Hypopotassemia Syndrome, Hypospadias-  
 Dysphagia Syndrome, Hyposmia, Hypothalamic Hamartoblastoma Hypopituitarism  
 Imperforate Anus Polydactyly, Hypothalamic Infantilism-Obesity, Hypothyroidism,  
 Hypotonia-Hypomentia-Hypogonadism-Obesity Syndrome, Hypoxanthine-Guanine  
 Phosphoribosyltransferase Defect (Complete Absence of), I-Cell Disease, Iatrogenic  
 10 Hypoglycemia, IBGC, IBIDS Syndrome, IBM, IBS, IC, I-Cell Disease, ICD, ICE  
 Syndrome Cogan-Reese Type, Icelandic Type Amyloidosis (Type VI), I-Cell Disease,  
 Ichthyosiform Erythroderma Corneal Involvement and Deafness, Ichthyosiform  
 Erythroderma Hair Abnormality Growth and Men, Ichthyosiform Erythroderma with  
 Leukocyte Vacuolation, Ichthyosis, Ichthyosis Congenita, Ichthyosis Congenital with  
 15 Trichothiodystrophy, Ichthyosis Hystrix, Ichthyosis Hystrix Gravior, Ichthyosis Linearis  
 Circumflexa, Ichthyosis Simplex, Ichthyosis Tay Syndrome, Ichthyosis Vulgaris,  
 Ichthyotic Neutral Lipid Storage Disease, Icteric Leptospirosis, Icterohemorrhagic  
 Leptospirosis, Icterus (Chronic Familial), Icterus Gravis Neonatorum, Icterus Intermittens  
 Juvenalis, Idiopathic Alveolar Hypoventilation, Idiopathic Amyloidosis, Idiopathic  
 20 Arteritis of Takayasu, Idiopathic Basal Ganglia Calcification (IBGC), Idiopathic Brachial  
 Plexus Neuropathy, Idiopathic Cervical Dystonia, Idiopathic Dilatation of the Pulmonary  
 Artery, Idiopathic Facial Palsy, Idiopathic Familial Hyperlipemia, Idiopathic Hypertrophic  
 Subaortic Stenosis, Idiopathic Hypoproteinemia, Idiopathic Immunoglobulin Deficiency,  
 Idiopathic Neonatal Hepatitis, Idiopathic Non-Specific Ulcerative Colitis, Idiopathic  
 25 Peripheral Periphlebitis, Idiopathic Pulmonary Fibrosis, Idiopathic Refractory  
 Sideroblastic Anemia, Idiopathic Renal Hematuria, Idiopathic Steatorrhea, Idiopathic  
 Thrombocythemia, Idiopathic Thrombocytopenic Purpura, Idiopathic Thrombocytopenia  
 Purpura (ITP), IDPA, IgA Nephropathy, IHSS, Ileitis, Ileocolitis, Illinois Type  
 Amyloidosis, ILS, IM, IMD2, IMD5, Immune Defect due to Absence of Thymus, Immune  
 30 Hemolytic Anemia Paroxysmal Cold, Immunodeficiency with Ataxia Telangiectasia,  
 Immunodeficiency Cellular with Abnormal Immunoglobulin Synthesis, Immunodeficiency

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Common Variable Unclassifiable, Immunodeficiency with Hyper-IgM, Immunodeficiency with Leukopenia, Immunodeficiency-2, Immunodeficiency-5 (IMD5), Immunoglobulin Deficiency, Imperforate Anus, Imperforate Anus with Hand Foot and Ear Anomalies, Imperforate Nasolacrimal Duct and Premature Aging Syndrome, Impotent Neutrophil

5 Syndrome, Inability To Open Mouth Completely And Short Finger-Flexor, INAD, Inborn Error of Urea Synthesis Arginase Type, Inborn Error of Urea Synthesis Arginino Succinic Type, Inborn Errors of Urea Synthesis Carbamyl Phosphate Type, Inborn Error of Urea Synthesis Citrullinemia Type, Inborn Errors of Urea Synthesis Glutamate Synthetase Type, INCL, Inclusion body myositis, Incomplete Atrioventricular Septal Defect, Incomplete

10 Testicular Feminization, Incontinentia Pigmenti, Incontinentia Pigmenti Achromians, Index Finger Anomaly with Pierre Robin Syndrome, Indiana Type Amyloidosis (Type II), Indolent systemic mastocytosis, Infantile Acquired Aphasia, Infantile Autosomal Recessive Polycystic Kidney Disease, Infantile Beriberi, Infantile Cerebral Ganglioside, Infantile Cerebral Paralysis, Infantile Cystinosis, Infantile Epileptic, Infantile Fanconi

15 Syndrome with Cystinosis, Infantile Finnish Type Neuronal Ceroid Lipofuscinosis, Infantile Gaucher Disease, Infantile Hypoglycemia, Infantile Hypophosphatasia, Infantile Lobar Emphysema, Infantile Myoclonic Encephalopathy, Infantile Myoclonic Encephalopathy and Polymyoclonia, Infantile Myofibromatosis, Infantile Necrotizing Encephalopathy, Infantile Neuronal Ceroid Lipofuscinosis, Infantile Neuroaxonal

20 Dystrophy, Infantile Onset Schindler Disease, Infantile Phytanic Acid Storage Disease, Infantile Refsum Disease (IRD), Infantile Sipoidosis GM-2 Gangliosidosis (Type S), Infantile Sleep Apnea, Infantile Spasms, Infantile Spinal Muscular Atrophy (all types), Infantile Spinal Muscular Atrophy ALS, Infantile Spinal Muscular Atrophy Type I, Infantile Type Neuronal Ceroid Lipofuscinosis, Infectious Jaundice, Inflammatory Breast

25 Cancer, Inflammatory Linear Nevus Sebaceous Syndrome, Iniencephaly, Insulin Resistant Acanthosis Nigricans, Insulin Lipodystrophy, Insulin dependent Diabetes, Intention Myoclonus, Intermediate Cystinosis, Intermediate Maple Syrup Urine Disease, Intermittent Ataxia with Pyruvate Dehydrogenase Deficiency, Intermittent Maple Syrup Urine Disease, Internal Hydrocephalus, Interstitial Cystitis, Interstitial Deletion of 4q Included, Intestinal

30 Lipodystrophy, Intestinal Lipophagic Granulomatosis, Intestinal Lymphangiectasia, Intestinal Polyposis I, Intestinal Polyposis II, Intestinal Polyposis III, Intestinal Polyposis-

Cutaneous Pigmentation Syndrome, Intestinal Pseudoobstruction with External  
 Ophthalmoplegia, Intracranial Neoplasm, Intracranial Tumors, Intracranial Vascular  
 Malformations, Intrauterine Dwarfism, Intrauterine Synechiae, Inverted Smile And Occult  
 Neuropathic Bladder, Iowa Type Amyloidosis (Type IV), IP, IPA, Iridocorneal Endothelial  
 5 Syndrome, Iridocorneal Endothelial (ICE) Syndrome Cogan-Resse Type,  
 Iridogoniodysgenesis With Somatic Anomalies, Iris Atrophy with Corneal Edema and  
 Glaucoma, Iris Nevus Syndrome, Iron Overload Anemia, Iron Overload Disease, Irritable  
 Bowel Syndrome, Irritable Colon Syndrome, Isaacs Syndrome, Isaacs-Merten Syndrome,  
 Ischemic Cardio myopathy, Isolated Lissencephaly Sequence, Isoleucine 33 Amyloidosis,  
 10 Isovaleric Acid CoA Dehydrogenase Deficiency, Isovaleric Acidaemia,  
 Isovalericacidemia, Isovaleryl CoA Carboxylase Deficiency, ITO Hypomelanosis, ITO,  
 ITP, IVA, Ivemark Syndrome, Iwanoff Cysts, Jackknife Convulsion, Jackson-Weiss  
 Craniosynostosis, Jackson-Weiss Syndrome, Jacksonian Epilepsy, Jacobsen Syndrome,  
 Jadassohn-Lewandowsky Syndrome, Jaffe-Lichenstein Disease, Jakob's Disease, Jakob-  
 15 Creutzfeldt Disease, Janeway I, Janeway Dysgammaglobulinemia, Jansen Metaphyseal  
 Dysostosis, Jansen Type Metaphyseal Chondrodysplasia, Jarcho-Levin Syndrome, Jaw-  
 Winking, JBS, JDMS, Jegher's Syndrome, Jejunal Atresia, Jejunitis, Jejunoileitis, Jervell  
 and Lange-Nielsen Syndrome, Jeune Syndrome, JMS, Job Syndrome, Job-Buckley  
 Syndrome, Johanson-Blizzard Syndrome, John Dalton, Johnson-Stevens Disease,  
 20 Jonston's Alopecia, Joseph's Disease, Joseph's Disease Type I, Joseph's Disease Type II,  
 Joseph's Disease Type III, Joubert Syndrome, Joubert-Bolthausen Syndrome, JRA, Juberg  
 Hayward Syndrome, Juberg-Marsidi Syndrome, Juberg-Marsidi Mental Retardation  
 Syndrome, Jumping Frenchmen, Jumping Frenchmen of Maine, Juvenile Arthritis,  
 Juvenile Autosomal Recessive Polycystic Kidney Disease, Juvenile Cystinosis, Juvenile  
 25 (Childhood) Dermatomyositis (JDMS), Juvenile Diabetes, Juvenile Gaucher Disease,  
 Juvenile Gout Choreoathetosis and Mental Retardation Syndrome, Juvenile Intestinal  
 Malabsorption of Vit B12, Juvenile Intestinal Malabsorption of Vitamin B12, Juvenile  
 Macular Degeneration, Juvenile Pernicious Anemia, Juvenile Retinoschisis, Juvenile  
 Rheumatoid Arthritis, Juvenile Spinal Muscular Atrophy Included, Juvenile Spinal  
 30 Muscular Atrophy ALS Included, Juvenile Spinal Muscular Atrophy Type III, Juxta-  
 Articular Adiposis Dolorosa, Juxtaglomerular Hyperplasia, Kabuki Make-Up Syndrome,

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Kahler Disease, Kallmann Syndrome, Kanner Syndrome, Kanzaki Disease, Kaposi Disease  
 (not Kaposi Sarcoma), Kappa Light Chain Deficiency, Karsch-Neugebauer Syndrome,  
 Kartagener Syndrome-Chronic Sinobronchial Disease and Dextrocardia, Kartagener Triad,  
 Kasabach-Merritt Syndrome, Kast Syndrome, Kawasaki Disease, Kawasaki Syndrome,  
 5 KBG Syndrome, KD, Kearns-Sayre Disease, Kearns-Sayre Syndrome, Kennedy Disease,  
 Kennedy Syndrome, Kennedy Type Spinal and Bulbar Muscular Atrophy, Kennedy-  
 Stefanis Disease, Kenny Disease, Kenny Syndrome, Kenny Type Tubular Stenosis,  
 Kenny-Caffe Syndrome, Kera. Palmoplant. Con. Pes Planus Ony. Periodon. Arach.,  
 Keratitis Ichthyosis Deafness Syndrome, Keratoconus, Keratoconus Posticus  
 10 Circumscriptus, Keratolysis, Keratolysis Exfoliativa Congenita, Keratolytic Winter  
 Erythema, Keratomalacia, Keratosis Follicularis, Keratosis Follicularis Spinulosa  
 Decalvans, Keratosis Follicularis Spinulosa Decalvans Ichthyosis, Keratosis Nigricans,  
 Keratosis Palmoplantaris with Periodontopathia and Onychogryposis, Keratosis  
 Palmoplantaris Congenital Pes Planus Onychogryposis Periodontosis Arachnodactyly,  
 15 Keratosis Palmoplantaris Congenital, Pes Planus, Onychogryphosis, Periodontosis,  
 Arachnodactyly, Acroosteolysis, Keratosis Rubra Figurata, Keratosis Seborrhoeica,  
 Ketoacid Decarboxylase Deficiency, Ketoaciduria, Ketotic Glycinemia, KFS, KID  
 Syndrome, Kidney Agenesis, Kidneys Cystic-Retinal Aplasia Joubert Syndrome, Killian  
 Syndrome, Killian/Teschler-Nicola Syndrome, Kiloh-Nevin syndrome III, Kinky Hair  
 20 Disease, Kinsbourne Syndrome, Kleeblattschadel Deformity, Kleine-Levin Syndrome,  
 Kleine-Levin Hibernation Syndrome, Klinefelter, Klippel-Feil Syndrome, Klippel-Feil  
 Syndrome Type I, Klippel-Feil Syndrome Type II, Klippel-Feil Syndrome Type III,  
 Klippel Trenaunay Syndrome, Klippel-Trenaunay-Weber Syndrome, Kluver-Bucy  
 Syndrome, KMS, Kniest Dysplasia, Kniest Syndrome, Kobner's Disease, Koebberling-  
 25 Dunnigan Syndrome, Kohlmeier-Degos Disease, Kok Disease, Korsakoff Psychosis,  
 Korsakoff's Syndrome, Krabbe's Disease Included, Krabbe's Leukodystrophy, Kramer  
 Syndrome, KSS, KTS, KTW Syndrome, Kufs Disease, Kugelberg-Welander Disease,  
 Kugelberg-Welander Syndrome, Kussmaul-Landry Paralysis, KWS, L-3-Hydroxy-Acyl-  
 CoA Dehydrogenase (LCHAD) Deficiency, Laband Syndrome, Labhart-Willi Syndrome,  
 30 Labyrinthine Syndrome, Labyrinthine Hydrops, Lacrimo-Auriculo-Dento-Digital  
 Syndrome, Lactase Isolated Intolerance, Lactase Deficiency, Lactation-Uterus Atrophy,

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Lactic Acidosis Leber Hereditary Optic Neuropathy, Lactic and Pyruvate Acidemia with  
 Carbohydrate Sensitivity, Lactic and Pyruvate Acidemia with Episodic Ataxia and  
 Weakness, Lactic and Pyruvate, Lactic acidosis, Lactose Intolerance of Adulthood,  
 Lactose Intolerance, Lactose Intolerance of Childhood, LADD Syndrome, LADD, Lafora  
 5 Disease Included, Lafora Body Disease, Laki-Lorand Factor Deficiency, LAM, Lambert  
 Type Ichthyosis, Lambert-Eaton Syndrome, Lambert-Eaton Myasthenic Syndrome,  
 Lamellar Recessive Ichthyosis, Lamellar Ichthyosis, Lancereaux-Mathieu-Weil  
 Spirochetosis, Landau-Kleffner Syndrome, Landouzy Dejerine Muscular Dystrophy,  
 Landry Ascending Paralysis, Langer-Salidino Type Achondrogensis (Type II), Langer  
 10 Giedion Syndrome, Langerhans-Cell Granulomatosis, Langerhans-Cell Histiocytosis  
 (LCH), Large Atrial and Ventricular Defect, Laron Dwarfism, Laron Type Pituitary  
 Dwarfism, Larsen Syndrome, Laryngeal Dystonia, Latah (Observed in Malaysia), Late  
 Infantile Neuroaxonal Dystrophy, Late Infantile Neuroaxonal Dystrophy, Late Onset  
 Cockayne Syndrome Type III (Type C), Late-Onset Dystonia, Late-Onset Immunoglobulin  
 15 Deficiency, Late Onset Pelizaeus-Merzbacher Brain Sclerosis, Lattice Corneal Dystrophy,  
 Lattice Dystrophy, Launois-Bensaude, Launois-Cleret Syndrome, Laurence Syndrome,  
 Laurence-Moon Syndrome, Laurence-Moon/Bardet-Biedl, Lawrence-Seip Syndrome,  
 LCA, LCAD Deficiency, LCAD, LCAD, LCADH Deficiency, LCH, LCHAD, LCPD, Le  
 Jeune Syndrome, Leband Syndrome, Leber's Amaurosis, Leber's Congenital  
 20 Amaurosis, Congenital Absence of the Rods and Cones, Leber's Congenital Tapetoretinal  
 Degeneration, Leber's Congenital Tapetoretinal Dysplasia, Leber's Disease, Leber's Optic  
 Atrophy, Leber's Optic Neuropathy, Left Ventricular Fibrosis, Leg Ulcer, Legg-Calve-  
 Perthes Disease, Leigh's Disease, Leigh's Syndrome, Leigh's Syndrome (Subacute  
 Necrotizing Encephalomyelopathy), Leigh Necrotizing Encephalopathy, Lennox-Gastaut  
 25 Syndrome, Lentigio-Polypose-Digestive Syndrome, Lenz Dysmorphogenetic Syndrome,  
 Lenz Dysplasia, Lenz Microphthalmia Syndrome, Lenz Syndrome, LEOPARD Syndrome,  
 Leprechaunism, Leptomenigeal Angiomatosis, Leptospiral Jaundice, Leri-Weill Disease,  
 Leri-Weil Dyschondrosteosis, Leri-Weil Syndrome, Lermoyez Syndrome, Leroy Disease,  
 Lesch Nyhan Syndrome, Lethal Infantile Cardio myopathy, Lethal Neonatal Dwarfism,  
 30 Lethal Osteochondrodysplasia, Letterer-Siwe Disease, Leukocytic Anomaly Albinism,  
 Leukocytic Inclusions with Platelet Abnormality, Leukodystrophy, Leukodystrophy with

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- Rosenthal Fibers, Leukoencephalitis Periaxialis Concentric, Levine-Critchley Syndrome, Levulosemia, Levy-Hollister Syndrome, LGMD, LGS, LHON, LIC, Lichen Ruber Acuminatus, Lichen Acuminatus, Lichen Amyloidosis, Lichen Planus, Lichen Psoriasis, Lignac-Debre-Fanconi Syndrome, Lignac-Fanconi Syndrome, Ligneous Conjunctivitis,
- 5 Limb-Girdle Muscular Dystrophy, Limb Malformations-Dento-Digital Syndrome, Limit Dextrinosis, Linear Nevroid Hypermelanosis, Linear Nevus Sebaceous Syndrome, Linear Scleroderma, Linear Sebaceous Nevus Sequence, Linear Sebaceous Nevus Syndrome, Lingua Fissurata, Lingua Plicata, Lingua Scrotalis, Linguofacial Dyskinesia, Lip Pseudocleft-hemangiomatic Branchial Cyst Syndrome, Lipid Granulomatosis, Lipid
- 10 Histiocytosis, Lipid Kerasin Type, Lipid Storage Disease, Lipid-Storage myopathy Associated with SCAD Deficiency, Lipidosis Ganglioside Infantile, Lipoatrophic Diabetes Mellitus, Lipodystrophy, Lipoid Corneal Dystrophy, Lipoid Hyperplasia-Male Pseudohermaphroditism, Lipomatosis of Pancreas Congenital, Lipomucopolysaccharidosis Type I, Lipomyelomeningocele, Lipoprotein Lipase Deficiency Familial, LIS, LIS1,
- 15 Lissencephaly 1, Lissencephaly Type I, Lissencephaly variants with agenesis of the corpus callosum cerebellar hypoplasia or other anomalies, Little Disease, Liver Phosphorylase Deficiency, LKS, LM Syndrome, Lobar Atrophy, Lobar Atrophy of the Brain, Lobar Holoprosencephaly, Lobar Tension Emphysema in Infancy, Lobstein Disease (Type I), Lobster Claw Deformity, Localized Epidermolysis Bullosa, Localized Lipodystrophy,
- 20 Localized Neuritis of the Shoulder Girdle, Loeffler's Disease, Loeffler Endomyocardial Fibrosis with Eosinophilia, Loeffler Fibroplastic Parietal Endocarditis, Loken Syndrome, Loken-Senior Syndrome, Long-Chain 3-hydroxyacyl-CoA Dehydrogenase (LCHAD), Long Chain Acyl CoA Dehydrogenase Deficiency, Long-Chain Acyl-CoA Dehydrogenase (ACADL), Long-Chain Acyl-CoA Dehydrogenase Deficiency, Long QT Syndrome
- 25 without Deafness, Lou Gehrig's Disease, Lou Gehrig's Disease Included, Louis-Bar Syndrome, Low Blood Sugar, Low-Density Beta Lipoprotein Deficiency, Low Imperforate Anus, Low Potassium Syndrome, Lowe syndrome, Lowe's Syndrome, Lowe-Bickel Syndrome, Lowe-Terry-MacLachlan Syndrome, LS, LTD, Lubs Syndrome, Luft Disease, Lumbar Canal Stenosis, Lumbar Spinal Stenosis, Lumbosacral Spinal Stenosis, Lundborg-
- 30 Unverricht Disease, Lundborg-Unverricht Disease Included, Lupus, Lupus, Lupus Erythematosus, Luschka-Magendie Foramina Atresia, Lyell Syndrome, Lyelles Syndrome,

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Lymphadenoid Goiter, Lymphangiectatic Protein-Losing Enteropathy, Lymphangioliomatosis, Lymphangioliomyomatosis, Lymphangiomas, Lymphatic Malformations, Lynch Syndromes, Lynch Syndrome I, Lynch Syndrome II, Lysosomal Alpha-N-Acetylgalactosaminidase Deficiency Schindler Type, Lysosomal Glycoaminoacid Storage Disease-Angiokeratoma Corporis Diffusum, Lysosomal Glucosidase Deficiency, MAA, Machado Disease, Machado-Joseph Disease, Macrencephaly, Macrocephaly, Macrocephaly Hemihypertrophy, Macrocephaly with Multiple Lipomas and Hemangiomas, Macrocephaly with Pseudopapilledema and Multiple Hemangiomas, Macroglobulinemia, Macroglossia, Macroglossia-Omphalocele

5 Visceromegaly Syndrome, Macrostomia Ablepharon Syndrome, Macrothrombocytopenia Familial Bernard-Soulier Type, Macula Lutea degeneration, Macular Amyloidosis, Macular Degeneration, Macular Degeneration Disciform, Macular Degeneration Senile, Macular Dystrophy, Macular Type Corneal Dystrophy, MAD, Madelung's Disease, Maffucci Syndrome, Major Epilepsy, Malabsorption, Malabsorption-Ectodermal

10 Dysplasia-Nasal Alar Hypoplasia, Maladie de Roger, Maladie de Tics, Male Malformation of Limbs and Kidneys, Male Turner Syndrome, Malignant Acanthosis, Malignant Acanthosis Nigricans, Malignant Astrocytoma, Malignant Atrophic Papulosis, Malignant Fever, Malignant Hyperphenylalaninemia, Malignant Hyperpyrexia, Malignant Hyperthermia, Malignant Melanoma, Malignant Tumors of the Central Nervous System,

15 Mallory-Weiss Laceration, Mallory-Weiss Tear, Mallory-Weiss Syndrome, Mammary Paget's Disease, Mandibular Ameloblastoma, Mandibulofacial Dysostosis, Mannosidosis, Map-Dot-Fingerprint Type Corneal Dystrophy, Maple Syrup Urine Disease, Marble Bones, Marchiafava-Micheli Syndrome, Marcus Gunn Jaw-Winking Syndrome, Marcus Gunn Phenomenon, Marcus Gunn Ptosis with jaw-winking, Marcus Gunn Syndrome,

20 Marcus Gunn (Jaw-Winking) Syndrome, Marcus Gunn Ptosis (with jaw-winking), Marden-Walker Syndrome, Marden-Walker Type Connective Tissue Disorder, Marfan's Abiotrophy, Marfan-Achard syndrome, Marfan Syndrome, Marfan's Syndrome I, Marfan's Variant, Marfanoid Hypermobility Syndrome, Marginal Corneal Dystrophy, Marie's Ataxia, Marie Disease, Marie-Sainton Disease, Marie Strumpell Disease, Marie-

25 Strumpell Spondylitis, Marinesco-Sjogren Syndrome, Marinesco-Sjogren-Gorland Syndrome, Marker X Syndrome, Maroteaux Lamy Syndrome, Maroteaux Type

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Acromesomelic Dysplasia, Marshall's Ectodermal Dysplasias With Ocular and Hearing Defects, Marshall-Smith Syndrome, Marshall Syndrome, Marshall Type Deafness-Myopia-Cataract-Saddle Nose, Martin-Albright Syndrome, Martin-Bell Syndrome, Martorell Syndrome, MASA Syndrome, Massive Myoclonia, Mast Cell Leukemia, Mastocytosis, Mastocytosis With an Associated Hematologic Disorder, Maumenee Corneal Dystrophy, Maxillary Ameloblastoma, Maxillofacial Dysostosis, Maxillonasal Dysplasia, Maxillonasal Dysplasia Binder Type, Maxillopalpebral Synkinesis, May-Hegglin Anomaly, MCAD Deficiency, MCAD, McArdle Disease, McCune-Albright, MCD, McKusick Type Metaphyseal Chondrodysplasia, MCR, MCTD, Meckel Syndrome, Meckel-Gruber Syndrome, Median Cleft Face Syndrome, Mediterranean Anemia, Medium-Chain Acyl-CoA dehydrogenase (ACADM), Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency, Medium-Chain Acyl-CoA Dehydrogenase Deficiency, Medullary Cystic Disease, Medullary Sponge Kidney, MEF, Megaesophagus, Megalencephaly, Megalencephaly with Hyaline Inclusion, Megalencephaly with Hyaline Panneuropathy, Megaloblastic Anemia, Megaloblastic Anemia of Pregnancy, Megalocornea-Mental Retardation Syndrome, Meier-Gorlin Syndrome, Meige's Lymphedema, Meige's Syndrome, Melanodermic Leukodystrophy, Melanoplakia-Intestinal Polyposis, Melanoplakia-Intestinal Polyposis, MELAS Syndrome, MELAS, Melkersson Syndrome, Melnick-Fraser Syndrome, Melnick-Needles Osteodysplasty, Melnick-Needles Syndrome, Membranous Lipodystrophy, Mendes Da Costa Syndrome, Meniere Disease, Ménière's Disease, Meningeal Capillary Angiomas, Menkes Disease, Menke's Syndrome I, Mental Retardation Aphasia Shuffling Gait Adducted Thumbs (MASA), Mental Retardation-Deafness-Skeletal Abnormalities-Coarse Face with Full Lips, Mental Retardation with Hypoplastic 5th Fingernails and Toenails, Mental Retardation with Osteocartilaginous Abnormalities, Mental Retardation-X-linked with Growth Delay-Deafness-Microgenitalism, Menzel Type OPCA, Mermaid Syndrome, MERRF, MERRF Syndrome, Merten-Singleton Syndrome, MES, Mesangial IGA Nephropathy, Mesenteric Lipodystrophy, Mesiodens-Cataract Syndrome, Mesodermal Dysmorphodystrophy, Mesomelic Dwarfism-Madelung Deformity, Metabolic Acidosis, Metachromatic Leukodystrophy, Metatarsus Varus, Metatropic Dwarfism Syndrome, Metatropic Dysplasia, Metatropic Dysplasia I, Metatropic Dysplasia II, Methylmalonic

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Acidemia, Methylmalonic Aciduria, Meulengracht's Disease, MFD1, MG, MH, MHA, Micrencephaly, Microcephalic Primordial Dwarfism I, Microcephaly, Microcephaly-Hiatal Hernia-Nephrosis Galloway Type, Microcephaly-Hiatal Hernia-Nephrotic Syndrome, Microcystic Corneal Dystrophy, Microcythemia, Microlissencephaly, Microphthalmia,

5 Microphthalmia or Anophthalmos with Associated Anomalies, Micropolygyria With Muscular Dystrophy, Microtia Absent Patellae Micrognathia Syndrome, Microvillus Inclusion Disease, MID, Midsystolic-click-late systolic murmur syndrome, Miescher's Type I Syndrome, Mikulicz Syndrome, Mikulicz-Radecki Syndrome, Mikulicz-Sjogren Syndrome, Mild Autosomal Recessive, Mild Intermediate Maple Syrup Urine Disease,

10 Mild Maple Syrup Urine Disease, Miller Syndrome, Miller-Dieker Syndrome, Miller-Fisher Syndrome, Milroy Disease, Minkowski-Chauffard Syndrome, Minor Epilepsy, Minot-Von Willebrand Disease, Mirror-Image Dextrocardia, Mitochondrial Beta-Oxidation Disorders, Mitochondrial and Cytosolic, Mitochondrial Cytopathy, Mitochondrial Cytopathy, Kearns-Sayre Type, Mitochondrial Encephalopathy,

15 Mitochondrial Encephalo myopathy Lactic Acidosis and Strokelike Episodes, Mitochondrial myopathy, Mitochondrial myopathy Encephalopathy Lactic Acidosis Stroke-Like Episode, Mitochondrial PEPCK Deficiency, Mitral-valve prolapse, Mixed Apnea, Mixed Connective Tissue Disease, Mixed Hepatic Porphyrria, Mixed Non-Fluent Aphasia, Mixed Sleep Apnea, Mixed Tonic and Clonic Torticollis, MJD, MKS, ML I, ML

20 II, ML III, ML IV, ML Disorder Type I, ML Disorder Type II, ML Disorder Type III, ML Disorder Type IV, MLNS, MMR Syndrome, MND, MNGIE, MNS, Mobitz I, Mobitz II, Mobius Syndrome, Moebius Syndrome, Moersch-Woltmann Syndrome, Mohr Syndrome, Monilethrix, Monomodal Visual Amnesia, Mononeuritis Multiplex, Mononeuritis Peripheral, Mononeuropathy Peripheral, Monosomy 3p2, Monosomy 9p Partial,

25 Monosomy 11q Partial, Monosomy 13q Partial, Monosomy 18q Syndrome, Monosomy X, Monostotic Fibrous Dysplasia, Morgagni-Turner-Albright Syndrome, Morphea, Morquio Disease, Morquio Syndrome, Morquio Syndrome A, Morquio Syndrome B, Morquio-Brailsford Syndrome, Morvan Disease, Mosaic Tetrasomy 9p, Motor Neuron Disease, Motor Neuron Syndrome, Motor Neurone Disease, Motoneuron Disease, Motoneurone

30 Disease, Motor System Disease (Focal and Slow), Moya-moya Disease, Moyamoya Disease, MPS, MPS I, MPS I H, MPS I H/S Hurler/Scheie Syndrome, MPS I S Scheie

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Syndrome, MPS II, MPS IIA, MPS IIB, MPS II-AR Autosomal Recessive Hunter Syndrome, MPS II-XR, MPS II-XR Severe Autosomal Recessive, MPS III, MPS III A B C and D Sanfilippo A, MPS IV, MPS IV A and B Morquio A, MPS V, MPS VI, MPS VI Severe Intermediate Mild Maroteaux-Lamy, MPS VII, MPS VII Sly Syndrome, MPS VIII,

5 MPS Disorder, MPS Disorder I, MPS Disorder II, MPS Disorder III, MPS Disorder VI, MPS Disorder Type VII, MRS, MS, MSA, MSD, MSL, MSS, MSUD, MSUD, MSUD Type Ib, MSUD Type II, Mucocutaneous Lymph Node Syndrome, Mucopolipidosis I, Mucopolipidosis II, Mucopolipidosis III, Mucopolipidosis IV, Mucopolysaccharidosis, Mucopolysaccharidosis I-H, Mucopolysaccharidosis I-S, Mucopolysaccharidosis II,

10 Mucopolysaccharidosis III, Mucopolysaccharidosis IV, Mucopolysaccharidosis VI, Mucopolysaccharidosis VII, Mucopolysaccharidosis Type I, Mucopolysaccharidosis Type II, Mucopolysaccharidosis Type III, Mucopolysaccharidosis Type VII, Mucosis, Mucosulfatidosis, Mucous Colitis, Mucoviscidosis, Mulibrey Dwarfism, Mulibrey Nanism Syndrome, Mullerian Duct Aplasia-Renal Aplasia-Cervicothoracic Somite Dysplasia,

15 Mullerian Duct-Renal-Cervicothoracic-Upper Limb Defects, Mullerian Duct and Renal Agenesis with Upper Limb and Rib Anomalies, Mullerian-Renal-Cervicothoracic Somite Abnormalities, Multi-Infarct Dementia Binswanger's Type, Multicentric Castleman's Disease, Multifocal Eosinophilic Granuloma, Multiple Acyl-CoA Dehydrogenase Deficiency, Multiple Acyl-CoA Dehydrogenase Deficiency / Glutaric Aciduria Type II,

20 Multiple Angiomas and Endochondromas, Multiple Carboxylase Deficiency, Multiple Cartilaginous Enchondroses, Multiple Cartilaginous Exostoses, Multiple Enchondromatosis, Multiple Endocrine Deficiency Syndrome Type II, Multiple Epiphyseal Dysplasia, Multiple Exostoses, Multiple Exostoses Syndrome, Multiple Familial Polyposis, Multiple Lentigines Syndrome, Multiple Myeloma, Multiple Neuritis

25 of the Shoulder Girdle, Multiple Osteochondromatosis, Multiple Peripheral Neuritis, Multiple Polyposis of the Colon, Multiple Pterygium Syndrome, Multiple Sclerosis, Multiple Sulfatase Deficiency, Multiple Symmetric Lipomatosis, Multiple System Atrophy, Multisynostotic Osteodysgenesis, Multisynostotic Osteodysgenesis with Long Bone Fractures, Mulvihill-Smith Syndrome, MURCS Association, Murk Jansen Type

30 Metaphyseal Chondrodysplasia, Muscle Carnitine Deficiency, Muscle Core Disease, Muscle Phosphofructokinase Deficiency, Muscular Central Core Disease, Muscular

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- Dystrophy, Muscular Dystrophy Classic X-linked Recessive, Muscular Dystrophy Congenital With Central Nervous System Involvement, Muscular Dystrophy Congenital Progressive with Mental Retardation, Muscular Dystrophy Facioscapulohumeral, Muscular Rheumatism, Muscular Rigidity - Progressive Spasm, Musculoskeletal Pain Syndrome,
- 5 Mutilating Acropathy, Mutism, mvp, MVP, MWS, Myasthenia Gravis, Myasthenia Gravis Pseudoparalytica, Myasthenic Syndrome of Lambert-Eaton, Myelinoclastic Diffuse Sclerosis, Myelomatosis, Myhre Syndrome, Myoclonic Astatic Petit Mal Epilepsy, Myoclonic Dystonia, Myoclonic Encephalopathy of Infants, Myoclonic Epilepsy, Myoclonic Epilepsy Hartung Type, Myoclonus Epilepsy Associated with Ragged Red
- 10 Fibers, Myoclonic Epilepsy and Ragged-Red Fiber Disease, Myoclonic Progressive Familial Epilepsy, Myoclonic Progressive Familial Epilepsy, Myoclonic Seizure, Myoclonus, Myoclonus Epilepsy, Myoencephalopathy Ragged-Red Fiber Disease, Myofibromatosis, Myofibromatosis Congenital, Myogenic Facio-Scapulo-Peroneal Syndrome, Myoneurogastrointestinal Disorder and Encephalopathy, Myopathic
- 15 Arthrogryposis Multiplex Congenita, Myopathic Carnitine Deficiency, Myopathy Central Fibrillar, myopathy Congenital Nonprogressive, myopathy Congenital Nonprogressive with Central Axis, myopathy with Deficiency of Carnitine Palmitoyltransferase, myopathy-Marinesco-Sjogren Syndrome, myopathy-Metabolic Carnitine Palmitoyltransferase Deficiency, myopathy Mitochondrial-Encephalopathy-Lactic
- 20 Acidosis-Stroke, myopathy with Sarcoplasmic Bodies and Intermediate Filaments, Myophosphorylase Deficiency, Myositis Ossificans Progressiva, Myotonia Atrophica, Myotonia Congenita, Myotonia Congenita Intermittens, Myotonic Dystrophy, Myotonic myopathy Dwarfism Chondrodystrophy Ocular and Facial Anomalies, Myotubular myopathy, Myotubular myopathy X-linked, Myproic Acid, Myriachit (Observed in
- 25 Siberia), Myxedema, N-Acetylglucosamine-1-Phosphotransferase Deficiency, N-Acetyl Glutamate Synthetase Deficiency, NADH-CoQ reductase deficiency, Naegeli Ectodermal Dysplasias, Nager Syndrome, Nager Acrofacial Dysostosis Syndrome, Nager Syndrome, NAGS Deficiency, Nail Dystrophy-Deafness Syndrome, Nail Dysgenesis and Hypodontia, Nail-Patella Syndrome, Nance-Horan Syndrome, Nanocephalic Dwarfism, Nanocephaly,
- 30 Nanophthalmia, Narcolepsy, Narcoleptic syndrome, NARP, Nasal-fronto-faciodyplasia, Nasal Alar Hypoplasia Hypothyroidism Pancreatic Achylia Congenital Deafness,

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- Nasomaxillary Hypoplasia, Nasu Lipodystrophy, NBIA1, ND, NDI, NDP, Necrotizing Encephalomyelopathy of Leigh's, Necrotizing Respiratory Granulomatosis, Neill-Dingwall Syndrome, Nelson Syndrome, Nemaline myopathy, Neonatal Adrenoleukodystrophy, Neonatal Adrenoleukodystrophy (NALD), Neonatal
- 5 Adrenoleukodystrophy (ALD), Neonatal Autosomal Recessive Polycystic Kidney Disease, Neonatal Dwarfism, Neonatal Hepatitis, Neonatal Hypoglycemia, Neonatal Lactose Intolerance, Neonatal Lymphedema due to Exudative Enteropathy, Neonatal Progeroid Syndrome, Neonatal Pseudo-Hydrocephalic Progeroid Syndrome of Wiedemann-Rautenstrauch, Neoplastic Arachnoiditis, Nephroblastom, Nephrogenic Diabetes Insipidus,
- 10 Nephronophthisis Familial Juvenile, Nephropathic Cystinosis, Nephropathy-Pseudohermaphroditism-Wilms Tumor, Nephrosis-Microcephaly Syndrome, Nephrosis-Neuronal Dismigration Syndrome, Nephrotic-Glycosuric-Dwarfism-Rickets-Hypophosphatemic Syndrome, Netherton Disease, Netherton Syndrome, Netherton Syndrome Ichthyosis, Nettleship Falls Syndrome (X-Linked), Neu-Laxova Syndrome,
- 15 Neuhauser Syndrome, Neural-tube defects, Neuralgic Amyotrophy, Neuraminidase Deficiency, Neuraocutaneous melanosis, Neurinoma of the Acoustic Nerve, Neurinoma, Neuroacanthocytosis, Neuroaxonal Dystrophy Schindler Type, Neurodegeneration with brain iron accumulation type 1 (NBIA1), Neurofibroma of the Acoustic Nerve, Neurogenic Arthrogryposis Multiplex Congenita, Neuromyelitis Optica, Neuromyotonia,
- 20 Neuromyotonia, Focal, Neuromyotonia, Generalized, Familial, Neuromyotonia, Generalized, Sporadic, Neuronal Axonal Dystrophy Schindler Type, Neuronal Ceroid Lipofuscinosis Adult Type, Neuronal Ceroid Lipofuscinosis Juvenile Type, Neuronal Ceroid Lipofuscinosis Type 1, Neuronopathic Acute Gaucher Disease, Neuropathic Amyloidosis, Neuropathic Beriberi, Neuropathy Ataxia and Retinitis Pigmentosa,
- 25 Neuropathy of Brachialplexus Syndrome, Neuropathy Hereditary Sensory Type I, Neuropathy Hereditary Sensory Type II, Neutral Lipid Storage Disease, Nevii, Nevroid Basal Cell Carcinoma Syndrome, Nevus, Nevus Cavernosus, Nevus Comedonicus, Nevus Depigmentosus, Nevus Sebaceous of Jadassohn, Nezelof's Syndrome, Nezelof's Thymic Aplasia, Nezelof Type Severe Combined Immunodeficiency, NF, NF1, NF2, NF-1, NF-2,
- 30 NHS, Niemann Pick Disease, Nieman Pick disease Type A (acute neuronopathic form), Nieman Pick disease Type B, Nieman Pick Disease Type C (chronic neuronopathic form),

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Nieman Pick disease Type D (Nova Scotia variant), Nieman Pick disease Type E, Nieman  
 Pick disease Type F (sea-blue histiocyte disease), Night Blindness, Nigrospinodontal  
 Degeneration, Niikawakuroki Syndrome, NLS, NM, Noack Syndrome Type I, Nocturnal  
 Myoclonus Hereditary Essential Myoclonus, Nodular Cornea Degeneration, Non-Bullous  
 5 CIE, Non-Bullous Congenital Ichthyosiform Erythroderma, Non-Communicating  
 Hydrocephalus, Non-Deletion Type Alpha-Thalassemia / Mental Retardation syndrome,  
 Non-Ketonic Hyperglycinemia Type I (NKH), Non-Ketotic Hyperglycinemia, Non-Lipid  
 Reticuloendotheliosis, Non-Neuronopathic Chronic Adult Gaucher Disease, Non-Scarring  
 Epidermolysis Bullosa, Nonarteriosclerotic Cerebral Calcifications, Nonarticular  
 10 Rheumatism, Noncerebral, Juvenile Gaucher Disease, Nondiabetic Glycosuria,  
 Nonischemic Cardio myopathy, Nonketotic Hypoglycemia and Carnitine Deficiency due  
 to MCAD Deficiency, Nonketotic Hypoglycemia Caused by Deficiency of Acyl-CoA  
 Dehydrogenase, Nonketotic Glycinemia, Nonne's Syndrome, Nonne-Milroy-Meige  
 Syndrome, Nonopalescent Opalescent Dentine, Nonpuerperal Galactorrhea-Amenorrhea,  
 15 Nonsecretory Myeloma, Nonspherocytic Hemolytic Anemia, Nontropical Sprue, Noonan  
 Syndrome, Norepinephrine, Normal Pressure Hydrocephalus, Norman-Roberts Syndrome,  
 Norrbottnian Gaucher Disease, Norrie Disease, Norwegian Type Hereditary Cholestasis,  
 NPD, NPS, NS, NSA, Nuchal Dystonia Dementia Syndrome, Nutritional Neuropathy,  
 Nyhan Syndrome, OAV Spectrum, Obstructive Apnea, Obstructive Hydrocephalus,  
 20 Obstructive Sleep Apnea, OCC Syndrome, Occlusive Thromboaropathy, OCCS, Occult  
 Intracranial Vascular Malformations, Occult Spinal Dysraphism Sequence, Ochoa  
 Syndrome, Ochronosis, Ochronotic Arthritis, OCR, OCRL, Octocephaly, Ocular Albinism,  
 Ocular Herpes, Ocular Myasthenia Gravis, Oculo-Auriculo-Vertebral Dysplasia, Oculo-  
 Auriculo-Vertebral Spectrum, Oculo-Bucco-Genital Syndrome, Oculocerebral Syndrome  
 25 with Hypopigmentation, Oculocerebrocutaneous Syndrome, Oculo-Cerebro-Renal,  
 Oculocerebrorenal Dystrophy, Oculocerebrorenal Syndrome, Oculocraniosomatic  
 Syndrome (obsolete), Oculocutaneous Albinism, Oculocutaneous Albinism Chediak-  
 Higashi Type, Oculo-Dento-Digital Dysplasia, Oculodentodigital Syndrome, Oculo-Dento-  
 Osseous Dysplasia, Oculo Gastrointestinal Muscular Dystrophy, Oculo Gastrointestinal  
 30 Muscular Dystrophy, Oculomandibulodyscephaly with hypotrichosis,  
 Oculomandibulofacial Syndrome, Oculomotor with Congenital Contractures and Muscle

- Atrophy, Oculosympathetic Palsy, ODD Syndrome, ODOD, Odontogenic Tumor, Odontotrichomelic Syndrome, OFD, OFD Syndrome, Ohio Type Amyloidosis (Type VII), OI, OI Congenita, OI Tarda, Oldfield Syndrome, Oligohydramnios Sequence, Oligophrenia Microphthalmos, Oligophrenic Polydystrophy, Olivopontocerebellar
- 5 Atrophy, Olivopontocerebellar Atrophy with Dementia and Extrapyrarnidal Signs, Olivopontocerebellar Atrophy with Retinal Degeneration, Olivopontocerebellar Atrophy I, Olivopontocerebellar Atrophy II, Olivopontocerebellar Atrophy III, Olivopontocerebellar Atrophy IV, Olivopontocerebellar Atrophy V, Ollier Disease, Ollier Osteochondromatosis, Omphalocele-Visceromegaly-Macroglossia Syndrome, Ondine's Curse, Onion-Bulb
- 10 Neuropathy, Onion Bulb Polyneuropathy, Onychosteodysplasia, Onychotrichodysplasia with Neutropenia, OPCA, OPCA I, OPCA II, OPCA III, OPCA IV, OPCA V, OPD Syndrome, OPD Syndrome Type I, OPD Syndrome Type II, OPD I Syndrome, OPD II Syndrome, Ophthalmoarthritis, Ophthalmoplegia-Intestinal Pseudoobstruction, Ophthalmoplegia, Pigmentary Degeneration of the Retina and Cardio myopathy,
- 15 Ophthalmoplegia Plus Syndrome, Ophthalmoplegia Syndrome, Opitz BBB Syndrome, Opitz BBB/G Compound Syndrome, Opitz BBBG Syndrome, Opitz-Frias Syndrome, Opitz G Syndrome, Opitz G/BBB Syndrome, Opitz Hypertelorism-Hypospadias Syndrome, Opitz-Kaveggia Syndrome, Opitz Oculogenitolaryngeal Syndrome, Opitz Trigenocephaly Syndrome, Opitz Syndrome, Opsoclonus, Opsoclonus-Myoclonus,
- 20 Ophthalmoneuromyelitis, Optic Atrophy Polyneuropathy and Deafness, Optic Neuroencephalomyelopathy, Optic Neuromyelitis, Opticomyelitis, Optochiasmatic Arachnoiditis, Oral-Facial Clefts, Oral-facial Dyskinesia, Oral Facial Dystonia, Oral-Facial-Digital Syndrome, Oral-Facial-Digital Syndrome Type I, Oral-Facial-Digital Syndrome I, Oral-Facial-Digital Syndrome II, Oral-Facial-Digital Syndrome III, Oral-
- 25 Facial-Digital Syndrome IV, Orbital Cyst with Cerebral and Focal Dermal Malformations, Ornithine Carbamyl Transferase Deficiency, Ornithine Transcarbamylase Deficiency, Orocraniodigital Syndrome, Orofaciodigital Syndrome, Oromandibular Dystonia, Orthostatic Hypotension, Osler-Weber-Rendu disease, Osseous-Oculo-Dento Dysplasia, Osseous-Oculo-Dento Dysplasia, Osteitis deformans, Osteochondrodystrophy Deformans,
- 30 Osteochondroplasia, Osteodysplasty of Melnick and Needles, Osteogenesis Imperfect, Osteogenesis Imperfecta, Osteogenesis Imperfecta Congenita, Osteogenesis Imperfecta

- Tarda, Osteohypertrophic Nevus Flammeus, Osteopathia Hyperostotica Scleroticans Multiplex Infantalis, Osteopathia Hyperostotica Scleroticans Multiplex Infantalis, Osteopathyrosis, Osteopetrosis, Osteopetrosis Autosomal Dominant Adult Type, Osteopetrosis Autosomal Recessive Malignant Infantile Type, Osteopetrosis Mild
- 5 Autosomal Recessive Intermediate Typ, Osteosclerosis Fragilis Generalisata, Osteosclerotic Myeloma, Ostium Primum Defect (endocardial cushion defects included), Ostium Secundum Defect, OTC Deficiency, Oto Palato Digital Syndrome, Oto-Palato-Digital Syndrome Type I, Oto-Palatal-Digital Syndrome Type II, Otodental Dysplasia, Otopalatodigital Syndrome, Otopalataldigital Syndrome Type II, Oudtshoorn Skin,
- 10 Ovarian Dwarfism Turner Type, Ovary Aplasia Turner Type, OWR, Oxalosis, Oxidase deficiency, Oxycephaly, Oxycephaly-Acrocephaly, P-V, PA, PAC, Pachyonychia Ichtyosiforme, Pachyonychia Congenita with Natal Teeth, Pachyonychia Congenita, Pachyonychia Congenita Keratosis Disseminata Circumscripta (follicularis), Pachyonychia Congenita Jadassohn-Lewandowsky Type, PAF with MSA, Paget's Disease, Paget's
- 15 Disease of Bone, Paget's Disease of the Breast, Paget's Disease of the Nipple, Paget's Disease of the Nipple and Areola, Pagon Syndrome, Painful Ophthalmoplegia, PAIS, Palatal Myoclonus, Palato-Oto-Digital Syndrome, Palatal-Oto-Digital Syndrome Type I, Palatal-Oto-Digital Syndrome Type II, Pallister Syndrome, Pallister-Hall Syndrome, Pallister-Killian Mosaic Syndrome, Pallister Mosaic Aneuploidy, Pallister Mosaic
- 20 Syndrome, Pallister Mosaic Syndrome Tetrasomy 12p, Pallister-W Syndrome, Palmoplantar Hyperkeratosis and Alopecia, Palsy, Pancreatic Fibrosis, Pancreatic Insufficiency and Bone Marrow Dysfunction, Pancreatic Ulcerogenic Tumor Syndrome, Panmyelophthisis, Panmyelopathy, Pantothenate kinase associated neurodegeneration (PKAN), Papillon-Lefevre Syndrome, Papillotonic Psuedotabes, Paralysis Periodica
- 25 Paramyotonia, Paralytic Beriberi, Paralytic Brachial Neuritis, Paramedian Lower Lip Pits-Popliteal Pyerygium Syndrome, Paramedian Diencephalic Syndrome, Paramyeloidosis, Paramyoclonus Multiple, Paramyotonia Congenita, Paramyotonia Congenita of Von Eulenburg, Parkinson's disease, Paroxysmal Atrial Tachycardia, Paroxysmal Cold Hemoglobinuria, Paroxysmal Dystonia, Paroxysmal Dystonia Choreathetosis, Paroxysmal
- 30 Kinesigenic Dystonia, Paroxysmal Nocturnal Hemoglobinuria, Paroxysmal Normal Hemoglobinuria, Paroxysmal Sleep, Parrot Syndrome, Parry Disease, Parry-Romberg

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Syndrome, Parsonage-Turner Syndrome, Partial Androgen Insensitivity Syndrome, Partial Deletion of the Short Arm of Chromosome 4, Partial Deletion of the Short Arm of Chromosome 5, Partial Deletion of Short Arm of Chromosome 9, Partial Duplication 3q Syndrome, Partial Duplication 15q Syndrome, Partial Facial Palsy With Urinary

5 Abnormalities, Partial Gigantism of Hands and Feet- Nevi-Hemihypertrophy-Macrocephaly, Partial Lipodystrophy, Partial Monosomy of Long Arm of Chromosome 11, Partial Monosomy of the Long Arm of Chromosome 13, Partial Spinal Sensory Syndrome, Partial Trisomy 11q, Parington Syndrome, PAT, Patent Ductus Arteriosus, Pathological Myoclonus, Pauciarticular-Onset Juvenile Arthritis, Paulitis, PBC, PBS, PC

10 Deficiency, PC Deficiency Group A, PC Deficiency Group B, PC, Eulenburg Disease, PCC Deficiency, PCH, PCLD, PCT, PD, PDA, PDH Deficiency, Pearson Syndrome Pyruvate Carboxylase Deficiency, Pediatric Obstructive Sleep Apnea, Peeling Skin Syndrome, Pelizaeus-Merzbacher Disease, Pelizaeus-Merzbacher Brain Sclerosis, Pellagra-Cerebellar Ataxia-Renal Aminoaciduria Syndrome, Pelvic Pain Syndrome,

15 Pemphigus Vulgaris, Pena Shokeir II Syndrome, Pena Shokeir Syndrome Type II, Penile Fibromatosis, Penile Fibrosis, Penile Induration, Penta X Syndrome, Pentalogy of Cantrell, Pentalogy Syndrome, Pentasomy X, PEPCK Deficiency, Pepper Syndrome, Perheentupa Syndrome, Periarticular Fibrositis, Pericardial Constriction with Growth Failure, Pericollagen Amyloidosis, Perinatal Polycystic Kidney Diseases, Perineal Anus, Periodic

20 Amyloid Syndrome, Periodic Peritonitis Syndrome, Periodic Somnolence and Morbid Hunger, Periodic Syndrome, Peripheral Cystoid Degeneration of the Retina, Peripheral Dysostosis-Nasal Hypoplasia-Mental Retardation, Peripheral Neuritis, Peripheral Neuropathy, Peritoneopericardial Diaphragmatic Hernia, Pernicious Anemia, Peromelia with Micrognathia, Peroneal Muscular Atrophy, Peroneal Nerve Palsy, Peroutka Sneeze,

25 Peroxisomal Acyl-CoA Oxidase, Peroxisomal Beta-Oxidation Disorders, Peroxisomal Bifunctional Enzyme, Peroxisomal Thiolase, Peroxisomal Thiolase Deficiency, Persistent Truncus Arteriosus, Perthes Disease, Petit Mal Epilepsy, Petit Mal Variant, Peutz-Jeghers Syndrome, Peutz-Touraine Syndrome, Peyronie Disease, Pfeiffer, Pfeiffer Syndrome Type I, PGA I, PGA II, PGA III, PGK, PH Type I, PH Type I, Pharyngeal Pouch Syndrome,

30 PHD Short-Chain Acyl-CoA Dehydrogenase Deficiency, Phenylalanine Hydroxylase Deficiency, Phenylalaninemia, Phenylketonuria, Phenylpyruvic Oligophrenia, Phocomelia,

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- Phocomelia Syndrome, Phosphoenolpyruvate Carboxykinase Deficiency, Phosphofructokinase Deficiency, Phosphoglycerate Kinase Deficiency, Phosphoglycerokinase, Phosphorylase 6 Kinase Deficiency, Phosphorylase Deficiency Glycogen Storage Disease, Phosphorylase Kinase Deficiency of Liver, Photic Sneeze
- 5 Reflex, Photoc Sneezing, Phototherapeutic keratectomy, PHS, Physicist John Dalton, Phytanic Acid Storage Disease, Pi Phenotype ZZ, PI, Pick Disease of the Brain, Pick's Disease, Pickwickian Syndrome, Pierre Robin Anomalad, Pierre Robin Complex, Pierre Robin Sequence, Pierre Robin Syndrome, Pierre Robin Syndrome with Hyperphalangy and Clinodactyly, Pierre-Marie's Disease, Pigmentary Degeneration of Globus Pallidus
- 10 Substantia Nigra Red Nucleus, Pili Torti and Nerve Deafness, Pili Torti-Sensorineural Hearing Loss, Pituitary Dwarfism II, Pituitary Tumor after Adrenalectomy, Pityriasis Pilaris, Pityriasis Rubra Pilaris, PJS, PKAN, PKD, PKD1, PKD2, PKD3, PKU, PKU1, Plagiocephaly, Plasma Cell Myeloma, Plasma Cell Leukemia, Plasma Thromboplastin Component Deficiency, Plasma Transglutaminase Deficiency, Plastic Induration Corpora
- 15 Cavemosa, Plastic Induration of the Penis, PLD, Plicated Tongue, PLS, PMD, Pneumorenal Syndrome, PNH, PNM, PNP Deficiency, POD, POH, Poikiloderma Atrophicans and Cataract, Poikiloderma Congenitale, Poland Anomaly, Poland Sequence, Poland Syndactyly, Poland Syndrome, Poliodystrophia Cerebri Progressiva, Polyarthritis Enterica, Polyarteritis Nodosa, Polyarticular-Onset Juvenile Arthritis Type I, Polyarticular-
- 20 Onset Juvenile Arthritis Type II, Polyarticular-Onset Juvenile Arthritis Types I and II, Polychondritis, Polycystic Kidney Disease, Polycystic Kidney Disease Medullary Type, Polycystic Liver Disease, Polycystic Ovary Disease, Polycystic Renal Diseases, Polydactyly-Joubert Syndrome, Polydysplastic Epidermolysis Bullosa, Polydytrophia Oligophrenia, Polydytrophic Dwarfism, Polyglandular Autoimmune Syndrome Type III,
- 25 Polyglandular Autoimmune Syndrome Type II, Polyglandular Autoimmune Syndrome Type I, Polyglandular Autoimmune Syndrome Type II, Polyglandular Deficiency Syndrome Type II, Polyglandular Syndromes, Polymorphic Macula Lutea Degeneration, Polymorphic Macular Degeneration, Polymorphism of Platelet Glycoprotein Ib, Polymorphous Corneal Dystrophy Hereditary, Polymyalgia Rheumatica, Polymyositis and
- 30 Dermatomyositis, Primary Agammaglobulinemia, Polyneuritis Peripheral, Polyneuropathy-Deafness-Optic Atrophy, Polyneuropathy Peripheral, Polyneuropathy and

- Polyradiculoneuropathy, Polyostotic Fibrous Dysplasia, Polyostotic Sclerosing  
 Histiocytosis, Polyposis Familial, Polyposis Gardner Type, Polyposis Hamartomatous  
 Intestinal, Polyposis-Osteomatosis-Epidermoid Cyst Syndrome, Polyposis Skin  
 Pigmentation Alopecia and Fingernail Changes, Polyps and Spots Syndrome, Polyserositis  
 5 Recurrent, Polysomy Y, Polysyndactyly with Peculiar Skull Shape, Polysyndactyly-  
 Dysmorphic Craniofacies Greig Type, Pompe Disease, Pompe Disease, Popliteal  
 Pterygium Syndrome, Porcupine Man, Porencephaly, Porencephaly, Porphobilinogen  
 deaminase (PBG-D), Porphyria, Porphyria Acute Intermittent, Porphyria ALA-D,  
 Porphyria Cutanea Tarda, Porphyria Cutanea Tarda Hereditaria, Porphyria Cutanea Tarda  
 10 Symptomatica, Porphyria Hepatica Variegata, Porphyria Swedish Type, Porphyria  
 Variegata, Porphyria Acute Intermittent, Porphyrins, Porriago Decalvans, Port Wine  
 Stains, Portuguese Type Amyloidosis, Post-Infective Polyneuritis, Postanoxic Intention  
 Myoclonus, Postaxial Acrofacial Dysostosis, Postaxial Polydactyly, Postencephalitic  
 Intention Myoclonus, Posterior Corneal Dystrophy Hereditary, Posterior Thalamic  
 15 Syndrome, Postmyelographic Arachnoiditis, Postnatal Cerebral Palsy, Postoperative  
 Cholestasis, Postpartum Galactorrhea-Amenorrhea Syndrome, Postpartum  
 Hypopituitarism, Postpartum Panhypopituitary Syndrome, Postpartum Panhypopituitarism,  
 Postpartum Pituitary Necrosis, Postural Hypotension, Potassium-Losing Nephritis,  
 Potassium Loss Syndrome, Potter Type I Infantile Polycystic Kidney Diseases, Potter Type  
 20 III Polycystic Kidney Disease, PPH, PPS, Prader-Willi Syndrome, Prader-Labhart-Willi  
 Fancone Syndrome, Prealbumin Tyr-77 Amyloidosis, Preexcitation Syndrome,  
 Pregnenolone Deficiency, Premature Atrial Contractions, Premature Senility Syndrome,  
 Premature Supraventricular Contractions, Premature Ventricular Complexes, Prenatal or  
 Connatal Neuroaxonal Dystrophy, Presenile Dementia, Presenile Macula Lutea Retinae  
 25 Degeneration, Primary Adrenal Insufficiency, Primary Agammaglobulinemias, Primary  
 Aldosteronism, Primary Alveolar Hypoventilation, Primary Amyloidosis, Primary Anemia,  
 Primary Beriberi, Primary Biliary, Primary Biliary Cirrhosis, Primary Brown Syndrome,  
 Primary Carnitine Deficiency, Primary Central Hypoventilation Syndrome, Primary  
 Ciliary Dyskinesia Kartagener Type, Primary Cutaneous Amyloidosis, Primary Dystonia,  
 30 Primary Failure Adrenocortical Insufficiency, Primary Familial Hypoplasia of the Maxilla,  
 Primary Hemochromatosis, Primary Hyperhidrosis, Primary Hyperoxaluria [Type I],

Primary Hyperoxaluria Type 1 (PH1), Primary Hyperoxaluria Type 1, Primary  
 Hyperoxaluria Type II, Primary Hyperoxaluria Type III, Primary Hypogonadism, Primary  
 Intestinal Lymphangiectasia, Primary Lateral Sclerosis, Primary Nonhereditary  
 Amyloidosis, Primary Obliterative Pulmonary Vascular Disease, Primary Progressive  
 5 Multiple Sclerosis, Primary Pulmonary Hypertension, Primary Reading Disability, Primary  
 Renal Glycosuria, Primary Sclerosing Cholangitis, Primary Thrombocythemia, Primary  
 Tumors of Central Nervous System, Primary Visual Agnosia, Proctocolitis Idiopathic,  
 Proctocolitis Idiopathic, Progeria of Adulthood, Progeria of Childhood, Progeroid Nanism,  
 Progeroid Short Stature with Pigmented Nevi, Progeroid Syndrome of De Barys,  
 10 Progressive Autonomic Failure with Multiple System Atrophy, Progressive Bulbar Palsy,  
 Progressive Bulbar Palsy Included, Progressive Cardiomyopathic Lentiginosis, Progressive  
 Cerebellar Ataxia Familial, Progressive Cerebral Poliodystrophy, Progressive Choroidal  
 Atrophy, Progressive Diaphyseal Dysplasia, Progressive Facial Hemiatrophy, Progressive  
 Familial Myoclonic Epilepsy, Progressive Hemifacial Atrophy, Progressive  
 15 Hypoerythemia, Progressive Infantile Poliodystrophy, Progressive Lenticular  
 Degeneration, Progressive Lipodystrophy, Progressive Muscular Dystrophy of Childhood,  
 Progressive Myoclonic Epilepsy, Progressive Osseous Heteroplasia, Progressive Pallid  
 Degeneration Syndrome, Progressive Spinobulbar Muscular Atrophy, Progressive  
 Supranuclear Palsy, Progressive Systemic Sclerosis, Progressive Tapetochoroidal  
 20 Dystrophy, Proline Oxidase Deficiency, Propionic Acidemia, Propionic Acidemia Type I  
 (PCCA Deficiency), Propionic Acidemia Type II (PCCB Deficiency), Propionyl CoA  
 Carboxylase Deficiency, Protanomaly, Protanopia, Protein-Losing Enteropathy Secondary  
 to Congestive Heart Failure, Proteus Syndrome, Proximal Deletion of 4q Included, PRP,  
 PRS, Prune Belly Syndrome, PS, Pseudo-Hurler Polydystrophy, Pseudo-Polydystrophy,  
 25 Pseudoacanthosis Nigricans, Pseudoachondroplasia, Pseudocholinesterase Deficiency,  
 Pseudogout Familial, Pseudohermophilia, Pseudohermaphroditism,  
 Pseudohermaphroditism-Nephron Disorder-Wilm's Tumor, Pseudohypertrophic Muscular  
 Dystrophy, Pseudohypoparathyroidism, Pseudohypophosphatasia, Pseudopolydystrophy,  
 Pseudothalidomide Syndrome, Pseudoxanthoma Elasticum, Psoriasis, Psorospermosis  
 30 Follicularis, PSP, PSS, Psychomotor Convulsion, Psychomotor Epilepsy, Psychomotor  
 Equivalent Epilepsy, PTC Deficiency, Pterygium, Pterygium Colli Syndrome, Pterygium

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Universale, Pterygolympangiectasia, Pulmonary Atresia, Pulmonary  
 Lymphangiomyomatosis, Pulmonary Stenosis, Pulmonic Stenosis-Ventricular Septal  
 Defect, Pulp Stones, Pulpal Dysplasia, Pulseless Disease, Pure A lymphocytosis, Pure  
 Cutaneous Histiocytosis, Purine Nucleoside Phosphorylase Deficiency, Purpura  
 5 Hemorrhagica, Purtilo Syndrome, PXE, PXE Dominant Type, PXE Recessive Type,  
 Pycnodysostosis, Pyknodysostosis, Pyknoepilepsy, Pyroglutamic Aciduria,  
 Pyroglutamicaciduria, Pyrroline Carboxylate Dehydrogenase Deficiency, Pyruvate  
 Carboxylase Deficiency, Pyruvate Carboxylase Deficiency Group A, Pyruvate  
 Carboxylase Deficiency Group B, Pyruvate Dehydrogenase Deficiency, Pyruvate Kinase  
 10 Deficiency, q25-qter, q26 or q27-qter, q31 or 32-qter, QT Prolongation with Extracellular  
 Hypohypocalcemia, QT Prolongation without Congenital Deafness, QT Prolonged with  
 Congenital Deafness, Quadriparesis of Cerebral Palsy, Quadriplegia of Cerebral Palsy,  
 Quantal Squander, Quantal Squander, r4, r6, r14, r 18, r21, r22, Rachischisis Posterior,  
 Radial Aplasia-Amegakaryocytic Thrombocytopenia, Radial Aplasia-Thrombocytopenia  
 15 Syndrome, Radial Nerve Palsy, Radicular Neuropathy Sensory, Radicular Neuropathy  
 Sensory Recessive, Radicular Dentin Dysplasia, Rapid-onset Dystonia-parkinsonism,  
 Rapp-Hodgkin Syndrome, Rapp-Hodgkin (hypohidrotic) Ectodermal Dysplasia syndrome,  
 Rapp-Hodgkin Hypohidrotic Ectodermal Dysplasias, Rare hereditary ataxia with  
 polyneuritic changes and deafness caused by a defect in the enzyme phytanic acid  
 20 hydroxylase, Rautenstrauch-Wiedemann Syndrome, Rautenstrauch-Wiedemann Type  
 Neonatal Progeria, Raynaud's Phenomenon, RDP, Reactive Functional Hypoglycemia,  
 Reactive Hypoglycemia Secondary to Mild Diabetes, Recessive Type Kenny-Caffé  
 Syndrome, Recklin Recessive Type Myotonia Congenita, Recklinghausen Disease,  
 Rectoperineal Fistula, Recurrent Vomiting, Reflex Neurovascular Dystrophy, Reflex  
 25 Sympathetic Dystrophy Syndrome, Refractive Errors, Refractory Anemia, Refrigeration  
 Palsy, Refsum Disease, Refsum's Disease, Regional Enteritis, Reid-Barlow's syndrome,  
 Reifenstein Syndrome, Reiger Anomaly-Growth Retardation, Reiger Syndrome, Reimann  
 Periodic Disease, Reimann's Syndrome, Reis-Bucklers Corneal Dystrophy, Reiter's  
 Syndrome, Relapsing Guillain-Barre Syndrome, Relapsing-Remitting Multiple Sclerosis,  
 30 Renal Agenesis, Renal Dysplasia-Blindness Hereditary, Renal Dysplasia-Retinal Aplasia  
 Loken-Senior Type, Renal Glycosuria, Renal Glycosuria Type A, Renal Glycosuria Type

B, Renal Glycosuria Type O, Renal-Oculocerebrodystrophy, Renal-Retinal Dysplasia with  
 Medullary Cystic Disease, Renal-Retinal Dystrophy Familial, Renal-Retinal Syndrome,  
 Rendu-Osler-Weber Syndrome, Respiratory Acidosis, Respiratory Chain Disorders,  
 Respiratory Myoclonus, Restless Legs Syndrome, Restrictive Cardio myopathy, Retention  
 5 Hyperlipemia, Rethore Syndrome (obsolete), Reticular Dysgenesis, Retinal Aplastic-  
 Cystic Kidneys-Joubert Syndrome, Retinal Cone Degeneration, Retinal Cone Dystrophy,  
 Retinal Cone-Rod Dystrophy, Retinitis Pigmentosa, Retinitis Pigmentosa and Congenital  
 Deafness, Retinoblastoma, Retinol Deficiency, Retinoschisis, Retinoschisis Juvenile,  
 Retraction Syndrome, Retrobulbar Neuropathy, Retrolenticular Syndrome, Rett Syndrome,  
 10 Reverse Coarction, Reye Syndrome, Reye's Syndrome, RGS, Rh Blood Factors, Rh  
 Disease, Rh Factor Incompatibility, Rh Incompatibility, Rhesus Incompatibility,  
 Rheumatic Fever, Rheumatoid Arthritis, Rheumatoid Myositis, Rhinosinusogenic Cerebral  
 Arachnoiditis, Rhizomelic Chondrodysplasia Punctata (RCDP), Acatlasemia, Classical  
 Refsum disease, RHS, Rhythmical Myoclonus, Rib Gap Defects with Micrognathia,  
 15 Ribbing Disease (obsolete), Ribbing Disease, Richner-Hanhart Syndrome, Rieger  
 Syndrome, Rieter's Syndrome, Right Ventricular Fibrosis, Riley-Day Syndrome, Riley-  
 Smith syndrome, Ring Chromosome 14, Ring Chromosome 18, Ring 4, Ring 4  
 Chromosome, Ring 6, Ring 6 Chromosome, Ring 9, Ring 9 Chromosome R9, Ring 14,  
 Ring 15, Ring 15 Chromosome (mosaic pattern), Ring 18, Ring Chromosome 18, Ring 21,  
 20 Ring 21 Chromosome, Ring 22, Ring 22 Chromosome, Ritter Disease, Ritter-Lyell  
 Syndrome, RLS, RMSS, Roberts SC-Phocomelia Syndrome, Roberts Syndrome, Roberts  
 Tetrachomelia Syndrome, Robertson's Ectodermal Dysplasias, Robin Anomalad, Robin  
 Sequence, Robin Syndrome, Robinow Dwarfism, Robinow Syndrome, Robinow  
 Syndrome Dominant Form, Robinow Syndrome Recessive Form, Rod myopathy, Roger  
 25 Disease, Rokitansky's Disease, Romano-Ward Syndrome, Romberg Syndrome, Rootless  
 Teeth, Rosenberg-Chutorian Syndrome, Rosewater Syndrome, Rosselli-Gulienatti  
 Syndrome, Rothmund-Thomson Syndrome, Roussy-Levy Syndrome, RP, RS X-Linked,  
 RS, RSDS, RSH Syndrome, RSS, RSTS, RTS, Rubella Congenital, Rubinstein Syndrome,  
 Rubinstein-Taybi Syndrome, Rubinstein Taybi Broad Thumb-Hallux syndrome, Rufous  
 30 Albinism, Ruhr's Syndrome, Russell's Diencephalic Cachexia, Russell's Syndrome,  
 Russell Syndrome, Russell-Silver Dwarfism, Russell-Silver Syndrome, Russell-Silver

- Syndrome X-linked, Ruvalcaba-Myhre-Smith syndrome (RMSS), Ruvalcaba Syndrome, Ruvalcaba Type Osseous Dysplasia with Mental Retardation, Sacral Regression, Sacral Agenesis Congenital, SAE, Saethre-Chotzen Syndrome, Sakati, Sakati Syndrome, Sakati-Nyhan Syndrome, Salaam Spasms, Salivosudoriparous Syndrome, Salzman Nodular
- 5 Corneal Dystrophy, Sandhoff Disease, Sanfilippo Syndrome, Sanfilippo Type A, Sanfilippo Type B, Santavuori Disease, Santavuori-Haltia Disease, Sarcoid of Boeck, Sarcoidosis, Sathre-chotzen, Saturday Night Palsy, SBMA, SC Phocomelia Syndrome, SC Syndrome, SCA 3, SCAD Deficiency, SCAD Deficiency Adult-Onset Localized, SCAD Deficiency Congenital Generalized, SCAD, SCADH Deficiency, Scalded Skin Syndrome,
- 10 Scalp Defect Congenital, Scaphocephaly, Scapula Elevata, Scapuloperoneal myopathy, Scapuloperoneal Muscular Dystrophy, Scapuloperoneal Syndrome Myopathic Type, Scarring Bullosa, SCHAD, Schaumann's Disease, Scheie Syndrome, Schereshevskii-Turner Syndrome, Schilder Disease, Schilder Encephalitis, Schilder's Disease, Schindler Disease Type I (Infantile Onset), Schindler Disease Infantile Onset, Schindler Disease, Schindler
- 15 Disease Type II (Adult Onset), Schinzel Syndrome, Schinzel-Giedion Syndrome, Schinzel Acrocallosal Syndrome, Schinzel-Giedion Midface-Retraktion Syndrome, Schizencephaly, Schmid Type Metaphyseal Chondrodysplasia, Schmid Metaphyseal Dysostosis, Schmid-Fraccaro Syndrome, Schmidt Syndrome, Schopf-Schultz-Passarge Syndrome, Schueller-Christian Disease, Schut-Haymaker Type, Schwartz-Jampel-Aberfeld Syndrome,
- 20 Schwartz-Jampel Syndrome Types 1A and 1B, Schwartz-Jampel Syndrome, Schwartz-Jampel Syndrome Type 2, SCID, Scleroderma, Sclerosis Familial Progressive Systemic, Sclerosis Diffuse Familial Brain, Scott Craniodigital Syndrome With Mental Retardation, Scrotal Tongue, SCS, SD, SDS, SDYS, Seasonal Conjunctivitis, Sebaceous Nevus Syndrome, Sebaceous nevus, Seborrhic Keratosis, Seborrhic Warts, Seckel Syndrome,
- 25 Seckel Type Dwarfism, Second Degree Congenital Heart Block, Secondary Amyloidosis, Secondary Blepharospasm, Secondary Non-tropical Sprue, Secondary Brown Syndrome, Secondary Beriberi, Secondary Generalized Amyloidosis, Secondary Dystonia, Secretory Component Deficiency, Secretory IgA Deficiency, SED Tarda, SED Congenital, SEDC, Segmental linear achromic nevus, Segmental Dystonia, Segmental Myoclonus, Seip
- 30 Syndrome, Seitelberger Disease, Seizures, Selective Deficiency of IgG Subclasses, Selective Mutism, Selective Deficiency of IgG Subclass, Selective IgM Deficiency,

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Selective Mutism, Selective IgA Deficiency, Self-Healing Histiocytosis, Semilobar Holoprosencephaly, Seminiferous Tubule Dysgenesis, Senile Retinoschisis, Senile Warts, Senior-Loken Syndrome, Sensory Neuropathy Hereditary Type I, Sensory Neuropathy Hereditary Type II, Sensory Neuropathy Hereditary Type I, Sensory Radicular

5 Neuropathy, Sensory Radicular Neuropathy Recessive, Septic Progressive Granulomatosis, Septo-Optic Dysplasia, Serous Circumscribed Meningitis, Serum Protease Inhibitor Deficiency, Serum Carnosinase Deficiency, Settleis Syndrome, Severe Combined Immunodeficiency, Severe Combined Immunodeficiency with Adenosine Deaminase Deficiency, Severe Combined Immunodeficiency (SCID), Sex Reversal, Sexual

10 Infantilism, SGB Syndrome, Sheehan Syndrome, Shields Type Dentinogenesis Imperfecta, Shingles, varicella-zoster virus, Ship Beriberi, SHORT Syndrome, Short Arm 18 Deletion Syndrome, Short Chain Acyl CoA Dehydrogenase Deficiency, Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency, Short Stature and Facial Telangiectasis, Short Stature Facial/Skeletal Anomalies-Retardation-Macrodonia, Short Stature-Hyperextensibility-

15 Rieger Anomaly-Teething Delay, Short Stature-Onychodysplasia, Short Stature Telangiectatic Erythema of the Face, SHORT Syndrome, Shoshin Beriberi, Shoulder girdle syndrome, Shprintzen-Goldberg Syndrome, Shulman Syndrome, Shwachman-Bodian Syndrome, Shwachman-Diamond Syndrome, Shwachman Syndrome, Shwachman-Diamond-Oski Syndrome, Shwachmann Syndrome, Shy Drager Syndrome, Shy-Magee

20 Syndrome, SI Deficiency, Sialidase Deficiency, Sialidosis Type I Juvenile, Sialidosis Type II Infantile, Sialidosis, Sialolipidosis, Sick Sinus Syndrome, Sickle Cell Anemia, Sickle Cell Disease, Sickle Cell-Hemoglobin C Disease, Sickle Cell-Hemoglobin D Disease, Sickle Cell-Thalassemia Disease, Sickle Cell Trait, Sideroblastic Anemias, Sideroblastic Anemia, Sideroblastosis, SIDS, Siegel-Cattan-Mamou Syndrome, Siemens-Bloch type

25 Pigmented Dermatoses, Siemens Syndrome, Siewerling-Creutzfeldt Disease, Siewert Syndrome, Silver Syndrome, Silver-Russell Dwarfism, Silver-Russell Syndrome, Simmond's Disease, Simons Syndrome, Simplex Epidermolysis Bullosa, Simpson Dysmorphia Syndrome, Simpson-Golabi-Behmel Syndrome, Sinding-Larsen-Johansson Disease, Singleton-Merten Syndrome, Sinus Arrhythmia, Sinus Venosus, Sinus

30 tachycardia, Sirenomelia Sequence, Sirenomelus, Situs Inversus Bronchiectasis and Sinusitis, SJA Syndrome, Sjogren Larsson Syndrome Ichthyosis, Sjogren Syndrome,

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Sjögren's Syndrome, SJS, Skeletal dysplasia, Skeletal Dysplasia Weismann Netter Stuhl  
 Type, Skin Peeling Syndrome, Skin Neoplasms, Skull Asymmetry and Mild Retardation,  
 Skull Asymmetry and Mild Syndactyly, SLE, Sleep Epilepsy, Sleep Apnea, SLO, Sly  
 Syndrome, SMA, SMA Infantile Acute Form, SMA I, SMA III, SMA type I, SMA type II,  
 5 SMA type III, SMA3, SMAX1, SMCR, Smith Lemli Opitz Syndrome, Smith Magenis  
 Syndrome, Smith-Magenis Chromosome Region, Smith-McCort Dwarfism, Smith-Opitz-  
 Inborn Syndrome, Smith Disease, Smoldering Myeloma, SMS, SNE, Sneezing From Light  
 Exposure, Sodium valproate, Solitary Plasmacytoma of Bone, Sorsby Disease, Sotos  
 Syndrome, Souques-Charcot Syndrome, South African Genetic Porphyria, Spasmodic  
 10 Dysphonia, Spasmodic Torticollis, Spasmodic Wryneck, Spastic Cerebral Palsy, Spastic  
 Colon, Spastic Dysphonia, Spastic Paraplegia, SPD Calcinosis, Specific Antibody  
 Deficiency with Normal Immunoglobulins, Specific Reading Disability, SPH2,  
 Spherocytic Anemia, Spherocytosis, Spherophakia-Brachymorphia Syndrome,  
 Sphingomyelin Lipidosis, Sphingomyelinase Deficiency, Spider fingers, Spielmeyer-Vogt  
 15 Disease, Spielmeyer-Vogt-Batten Syndrome, Spina Bifida, Spina Bifida Aperta, Spinal  
 Arachnoiditis, Spinal Arteriovenous Malformation, Spinal Ataxia Hereditofamilial, Spinal  
 and Bulbar Muscular Atrophy, Spinal Diffuse Idiopathic Skeletal Hyperostosis, Spinal  
 DISH, Spinal Muscular Atrophy, Spinal Muscular Atrophy All Types, Spinal Muscular  
 Atrophy Type ALS, Spinal Muscular Atrophy-Hypertrophy of the Calves, Spinal Muscular  
 20 Atrophy Type I, Spinal Muscular Atrophy Type III, Spinal Muscular Atrophy type 3,  
 Spinal Muscular Atrophy-Hypertrophy of the Calves, Spinal Ossifying Arachnoiditis,  
 Spinal Stenosis, Spino Cerebellar Ataxia, Spinocerebellar Atrophy Type I, Spinocerebellar  
 Ataxia Type I (SCA1), Spinocerebellar Ataxia Type II (SCAII), Spinocerebellar Ataxia  
 Type III (SCAIII), Spinocerebellar Ataxia Type III (SCA 3), Spinocerebellar Ataxia Type  
 25 IV (SCAIV), Spinocerebellar Ataxia Type V (SCAV), Spinocerebellar Ataxia Type VI  
 (SCAVI), Spinocerebellar Ataxia Type VII (SCAVII), Spirochetal Jaundice, Splenic  
 Agenesis Syndrome, Splenic Ptosis, Splenoptosis, Split Hand Deformity-Mandibulofacial  
 Dysostosis, Split Hand Deformity, Spondyloarthritis, Spondylocostal Dysplasia - Type I,  
 Spondyloepiphyseal Dysplasia Tarda, Spondylothoracic Dysplasia, Spondylotic Caudal  
 30 Radiculopathy, Sponge Kidney, Spongioblastoma Multiforme, Spontaneous  
 Hypoglycemia, Sprengel Deformity, Spring Ophthalmia, SRS, ST, Stale Fish Syndrome,

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Staphylococcal Scalded Skin Syndrome, Stargardt's Disease, Startle Disease, Status Epilepticus, Steele-Richardson-Olszewski Syndrome, Steely Hair Disease, Stein-Leventhal Syndrome, Steinert Disease, Stengel's Syndrome, Stengel-Batten-Mayou-Spielmeyer-Vogt-Stock Disease, Stenosing Cholangitis, Stenosis of the Lumbar Vertebral Canal, 5 Stenosis, Steroid Sulfatase Deficiency, Stevanovic's Ectodermal Dysplasias, Stevens Johnson Syndrome, STGD, Stickler Syndrome, Stiff-Man Syndrome, Stiff Person Syndrome, Still's Disease, Stilling-Turk-Duane Syndrome, Stillis Disease, Stimulus-Sensitive Myoclonus, Stone Man Syndrome, Stone Man, Streeter Anomaly, Striatonigral Degeneration Autosomal Dominant Type, Striopallidodentate Calcinosi, Stroma, 10 Descemet's Membrane, Stromal Corneal Dystrophy, Struma Lymphomatosa, Sturge-Kalischer-Weber Syndrome, Sturge Weber Syndrome, Sturge-Weber Phakomatosis, Subacute Necrotizing Encephalomyelopathy, Subacute Spongiform Encephalopathy, Subacute Necrotizing Encephalopathy, Subacute Sarcoidosis, Subacute Neuronopathic, Subaortic Stenosis, Subcortical Arteriosclerotic Encephalopathy, Subendocardial Sclerosis, 15 Succinylcholine Sensitivity, Sucrase-Isomaltase Deficiency Congenital, Sucrose-Isomaltose Malabsorption Congenital, Sucrose Intolerance Congenital, Sudanophilic Leukodystrophy ADL, Sudanophilic Leukodystrophy Pelizaeus-Merzbacher Type, Sudanophilic Leukodystrophy Included, Sudden Infant Death Syndrome, Sudeck's Atrophy, Sugio-Kajii Syndrome, Summerskill Syndrome, Summit Acrocephalosyndactyly, 20 Summitt's Acrocephalosyndactyly, Summitt Syndrome, Superior Oblique Tendon Sheath Syndrome, Suprarenal glands, Supravalvular Aortic Stenosis, Supraventricular tachycardia, Surdicardiac Syndrome, Surdocardiac Syndrome, SVT, Sweat Gland Abscess, Sweating Gustatory Syndrome, Sweet Syndrome, Swiss Cheese Cartilage Syndrome, Syndactylic Oxycephaly, Syndactyly Type I with Microcephaly and Mental Retardation, Syndromatic 25 Hepatic Ductular Hypoplasia, Syringomyelia, Systemic Aleukemic Reticuloendotheliosis, Systemic Amyloidosis, Systemic Carnitine Deficiency, Systemic Elastorrhexis, Systemic Lupus Erythematosus, Systemic Mast Cell Disease, Systemic Mastocytosis, Systemic-Onset Juvenile Arthritis, Systemic Sclerosis, Systopic Spleen, T-Lymphocyte Deficiency, Tachyalimentation Hypoglycemia, Tachycardia, Takahara syndrome, Takayasu Disease, 30 Takayasu Arteritis, Talipes Calcaneus, Talipes Equinovarus, Talipes Equinus, Talipes Varus, Talipes Valgus, Tandem Spinal Stenosis, Tangier Disease, Tapetoretinal

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Degeneration, TAR Syndrome, Tardive Dystonia, Tardive Muscular Dystrophy, Tardive  
 Dyskinesia, Tardive Oral Dyskinesia, Tardive Dystonia, Tardy Ulnar Palsy, Target Cell  
 Anemia, Tarsomegaly, Tarui Disease, TAS Midline Defects Included, TAS Midline  
 Defect, Tay Sachs Sphingolipidosis, Tay Sachs Disease, Tay Syndrome Ichthyosis, Tay  
 5 Sachs Sphingolipidosis, Tay Syndrome Ichthyosis, Taybi Syndrome Type I, Taybi  
 Syndrome, TCD, TCOF1, TCS, TD, TDO Syndrome, TDO-I, TDO-II, TDO-III,  
 Telangiectasis, Telecanthus with Associated Abnormalities, Telecanthus-Hypospadias  
 Syndrome, Temporal Lobe Epilepsy, Temporal Arteritis/Giant Cell Arteritis, Temporal  
 Arteritis, TEN, Tendon Sheath Adherence Superior Obliqu, Tension Myalgia, Terminal  
 10 Deletion of 4q Included, Terrian Corneal Dystrophy, Teschler-Nicola/Killian Syndrome,  
 Tethered Spinal Cord Syndrome, Tethered Cord Malformation Sequence, Tethered Cord  
 Syndrome, Tethered Cervical Spinal Cord Syndrome, Tetrahydrobiopterin Deficiencies,  
 Tetrahydrobiopterin Deficiencies, Tetralogy of Fallot, Tetraphocomelia-Thrombocytopenia  
 Syndrome, Tetrasomy Short Arm of Chromosome 9, Tetrasomy 9p, Tetrasomy Short Arm  
 15 of Chromosome 18, Thalamic Syndrome, Thalamic Pain Syndrome, Thalamic  
 Hyperesthetic Anesthesia, Thalassemia Intermedia, Thalassemia Minor, Thalassemia  
 Major, Thiamine Deficiency, Thiamine-Responsive Maple Syrup Urine Disease, Thin-  
 Basement-Membrane Nephropathy, Thiolase deficiency, RCDP, Acyl-CoA  
 dihydroxyacetonephosphate acyltransferase, Third and Fourth Pharyngeal Pouch  
 20 Syndrome, Third Degree Congenital (Complete) Heart Block, Thomsen Disease, Thoracic-  
 Pelvic-Phalangeal Dystrophy, Thoracic Spinal Canal, Thoracoabdominal Syndrome,  
 Thoracoabdominal Ectopia Cordis Syndrome, Three M Syndrome, Three-M Slender-  
 Boned Nanism, Thrombasthenia of Glanzmann and Naegeli, Thrombocythemia Essential,  
 Thrombocytopenia-Absent Radius Syndrome, Thrombocytopenia-Hemangioma  
 25 Syndrome, Thrombocytopenia-Absent Radii Syndrome, Thrombophilia Hereditary Due to  
 AT III, Thrombotic Thrombocytopenic Purpura, Thrombocytopenic Colitis, Thymic  
 Dysplasia with Normal Immunoglobulins, Thymic Agenesis, Thymic Aplasia DiGeorge  
 Type, Thymic Hypoplasia Agammaglobulinemias Primary Included, Thymic Hypoplasia  
 DiGeorge Type, Thymus Congenital Aplasia, Tic Douloureux, Tics, Tinel's syndrome,  
 30 Tolosa Hunt Syndrome, Tonic Spasmodic Torticollis, Tonic Pupil Syndrome, Tooth and  
 Nail Syndrome, Torch Infection, TORCH Syndrome, Torsion Dystonia, Torticollis, Total

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Lipodystrophy, Total anomalous pulmonary venous connection, Touraine's Aphthosis,  
 Tourette Syndrome, Tourette's disorder, Townes-Brocks Syndrome, Townes Syndrome,  
 Toxic Paralytic Anemia, Toxic Epidermal Necrolysis, Toxopachyosteose Diaphysaire  
 Tibio-Peroniere, Toxopachyosteose, Toxoplasmosis Other Agents Rubella  
 5 Cytomegalovirus Herpes Simplex, Tracheoesophageal Fistula with or without Esophageal  
 Atresia, Tracheoesophageal Fistula, Transient neonatal myasthenia gravis, Transitional  
 Atrioventricular Septal Defect, Transposition of the great arteries, Transtelephonic  
 Monitoring, Transthyretin Methionine-30 Amyloidosis (Type I), Trapezoidocephaly-  
 Multiple Synostosis Syndrome, Treacher Collins Syndrome, Treacher Collins-  
 10 Franceschetti Syndrome 1, Trevor Disease, Triatrial Heart, Tricho-Dento-Osseous  
 Syndrome, Trichodento Osseous Syndrome, Trichopoliodystrophy, Trichorhinophalangeal  
 Syndrome, Trichorhinophalangeal Syndrome, Tricuspid atresia, Trifunctional Protein  
 Deficiency, Trigeminal Neuralgia, Triglyceride Storage Disease Impaired Long-Chain  
 Fatty Acid Oxidation, Trigonitis, Trigonocephaly, Trigonocephaly Syndrome,  
 15 Trigonocephaly "C" Syndrome, Trimethylaminuria, Triphalangeal Thumbs-Hypoplastic  
 Distal Phalanges-Onychodystrophy, Triphalangeal Thumb Syndrome, Triple Symptom  
 Complex of Behcet, Triple X Syndrome, Triplo X Syndrome, Triploid Syndrome,  
 Triploidy, Triploidy Syndrome, Trismus-Pseudocamptodactyly Syndrome, Trisomy,  
 Trisomy G Syndrome, Trisomy X, Trisomy 6q Partial, Trisomy 6q Syndrome Partial,  
 20 Trisomy 9 Mosaic, Trisomy 9P Syndrome (Partial) Included, Trisomy 11q Partial, Trisomy  
 14 Mosaic, Trisomy 14 Mosaicism Syndrome, Trisomy 21 Syndrome, Trisomy 22 Mosaic,  
 Trisomy 22 Mosaicism Syndrome, TRPS, TRPS1, TRPS2, TRPS3, True  
 Hermaphroditism, Truncus arteriosus, Tryptophan Malabsorption, Tryptophan Pyrrolase  
 Deficiency, TS, TTP, TTTS, Tuberous Sclerosis, Tubular Ectasia, Turcot Syndrome,  
 25 Turner Syndrome, Turner-Kieser Syndrome, Turner Phenotype with Normal  
 Chromosomes (Karyotype), Turner-Varny Syndrome, Turricephaly, Twin-Twin  
 Transfusion Syndrome, Twin-to-Twin Transfusion Syndrome, Type A, Type B, Type AB,  
 Type O, Type I Diabetes, Type I Familial Incomplete Male, Type I Familial Incomplete  
 Male Pseudohermaphroditism, Type I Gaucher Disease, Type I (PCCA Deficiency), Type  
 30 I Tyrosinemia, Type II Gaucher Disease, Type II Histiocytosis, Type II (PCCB  
 Deficiency), Type II Tyrosinemia, Type IIA Distal Arthrogryposis Multiplex Congenita,

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Type III Gaucher Disease, Type III Tyrosinemia, Type III Dentinogenesis Imperfecta, Typical Retinoschisis, Tyrosinase Negative Albinism (Type I), Tyrosinase Positive Albinism (Type II), Tyrosinemia type 1 acute form, Tyrosinemia type 1 chronic form, Tyrosinosis, UCE, Ulcerative Colitis, Ulcerative Colitis Chronic Non-Specific, Ulnar-Mammary Syndrome, Ulnar-Mammary Syndrome of Pallister, Ulnar Nerve Palsy, UMS, Unclassified FODs, Unconjugated Benign Bilirubinemia, Underactivity of Parathyroid, Unilateral Ichthyosiform Erythroderma with Ipsilateral Malformations Limb, Unilateral Chondromatosis, Unilateral Defect of Pectoralis Muscle and Syndactyly of the Hand, Unilateral Hemidysplasia Type, Unilateral Megalencephaly, Unilateral Partial Lipodystrophy, Unilateral Renal Agenesis, Unstable Colon, Unverricht Disease, Unverricht-Lundborg Disease, Unverricht-Lundborg-Laf Disease, Unverricht Syndrome, Upper Limb - Cardiovascular Syndrome (Holt-Oram), Upper Motor Neuron Disease, Upper Airway Apnea, Urea Cycle Defects or Disorders, Urea Cycle Disorder Arginase Type, Urea Cycle Disorder Arginino Succinase Type, Urea Cycle Disorders Carbamyl Phosphate Synthetase Type, Urea Cycle Disorder Citrullinemia Type, Urea Cycle Disorders N-Acetyl Glutamate Synthetase Type, Urea Cycle Disorder OTC Type, Urethral Syndrome, Urethro-Oculo-Articular Syndrome, Uridine Diphosphate Glucuronosyltransferase Severe Def. Type I, Urinary Tract Defects, Urofacial Syndrome, Uroporphyrinogen III cosynthase, Urticaria pigmentosa, Usher Syndrome, Usher Type I, Usher Type II, Usher Type III, Usher Type IV, Uterine Synechiae, Uroporphyrinogen I-synthase, Uveitis, Uveomeningitis Syndrome, V-CJD, VACTERL Association, VACTERL Association, VACTERL Syndrome, Valgus Calcaneus, Valine Transaminase Deficiency, Valinemia, Valproic Acid, Valproate acid exposure, Valproic acid exposure, Valproic acid, Van Buren's Disease, Van der Hoeve-Habertsma-Waardenburg-Gauldi Syndrome, Variable Onset Immunoglobulin Deficiency Dysgammaglobulinemia, Variant Creutzfeldt-Jakob Disease (V-CJD), Varicella Embryopathy, Variegate Porphyria, Vascular Birthmarks, Vascular Dementia Binswanger's Type, Vascular Erectile Tumor, Vascular Hemophilia, Vascular Malformations, Vascular Malformations of the Brain, Vasculitis, Vasomotor Ataxia, Vasopressin-Resistant Diabetes Insipidus, Vasopressin-Sensitive Diabetes Insipidus, VATER Association, Vcf syndrome, Vcfs, Velocardiofacial Syndrome, VeloCardioFacial Syndrome, Venereal Arthritis, Venous Malformations, Ventricular

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Fibrillation, Ventricular Septal Defects, Congenital Ventricular Defects, Ventricular Septal Defect, Ventricular Tachycardia, Venual Malformations, VEOHD, Vermis Aplasia, Vermis Cerebellar Agenesis, Vernal Keratoconjunctivitis, Verruca, Vertebral Anal Tracheoesophageal Esophageal Radial, Vertebral Ankylosing Hyperostosis, Very Early  
 5 Onset Huntington's Disease, Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency, Vestibular Schwannoma, Vestibular Schwannoma Neurofibromatosis, Vestibulocerebellar, Virchow's Oxycephaly, Visceral Xanthogranulomatosis, Visceral Xantho-Granulomatosis, Visceral myopathy-External Ophthalmoplegia, Visceromegaly-Umbilical Hernia-Macroglossia Syndrome, Visual Amnesia, Vitamin A Deficiency,  
 10 Vitamin B-1 Deficiency, Vitelline Macular Dystrophy, Vitiligo, Vitiligo Capitis, Vitreoretinal Dystrophy, VKC, VKH Syndrome, VLCAD, Vogt Syndrome, Vogt Cephalosyndactyly, Vogt Koyanagi Harada Syndrome, Von Bechterew-Strumpell Syndrome, Von Eulenburg Paramyotonia Congenita, Von Frey's Syndrome, Von Gierke Disease, Von Hippel-Lindau Syndrome, Von Mikulicz Syndrome, Von Recklinghausen  
 15 Disease, Von Willebrandt Disease, VP, Vrolik Disease (Type II), VSD, Vulgaris Type Disorder of Cornification, Vulgaris Type Ichthyosis, W Syndrome, Waardenburg Syndrome, Waardenburg-Klein Syndrome, Waardenburg Syndrome Type I (WS1), Waardenburg Syndrome Type II (WS2), Waardenburg Syndrome Type IIA (WS2A), Waardenburg Syndrome Type IIB (WS2B), Waardenburg Syndrome Type III (WS3),  
 20 Waardenburg Syndrome Type IV (WS4), Waelsch's Syndrome, WAGR Complex, WAGR Syndrome, Waldenstroem's Macroglobulinemia, Waldenstrom's Purpura, Waldenstrom's Syndrome, Waldmann Disease, Walker-Warburg Syndrome, Wandering Spleen, Warburg Syndrome, Warm Antibody Hemolytic Anemia, Warm Reacting Antibody Disease, Wartenberg Syndrome, WAS, Water on the Brain, Watson Syndrome, Watson-Alagille  
 25 Syndrome, Waterhouse-Friderichsen syndrome, Waxy Disease, WBS, Weaver Syndrome, Weaver-Smith Syndrome, Weber-Cockayne Disease, Wegener's Granulomatosis, Weil Disease, Weil Syndrome, Weill-Marchesani, Weill-Marchesani Syndrome, Weill-Reyes Syndrome, Weismann-Netter-Stuhl Syndrome, Weissenbacher-Zweymuller Syndrome, Wells Syndrome, Wenckebach, Werdnig-Hoffman Disease, Werdnig-Hoffman Paralysis,  
 30 Werlhof's Disease, Werner Syndrome, Wernicke's (C) I Syndrome, Wernicke's aphasia, Wernicke-Korsakoff Syndrome, West Syndrome, Wet Beriberi, WHCR, Whipple's

Disease, Whipple Disease, Whistling face syndrome, Whistling Face-Windmill Vane Hand Syndrome, White-Darier Disease, Whitnall-Norman Syndrome, Whorled nevoid hypermelanosis, WHS, Wieacker Syndrome, Wieacher Syndrome, Wieacker-Wolff Syndrome, Wiedmann-Beckwith Syndrome, Wiedemann-Rautenstrauch Syndrome, Wildervanck Syndrome, Willebrand-Juergens Disease, Willi-Prader Syndrome, Williams Syndrome, Williams-Beuren Syndrome, Wilms' Tumor, Wilms' Tumor-Aniridia-Gonadoblastoma-Mental Retardation Syndrome, Wilms Tumor Aniridia Gonadoblastoma Mental Retardation, Wilms' Tumor-Aniridia-Genitourinary Anomalies-Mental Retardation Syndrome, Wilms Tumor-Pseudohermaphroditism-Nephropathy, Wilms Tumor and Pseudohermaphroditism, Wilms Tumor-Pseudohermaphroditism-Glomerulopathy, Wilson's Disease, Winchester Syndrome, Winchester-Grossman Syndrome, Wiskott-Aldrich Syndrome, Wiskott-Aldrich Type Immunodeficiency, Witkop Ectodermal Dysplasias, Witkop Tooth-Nail Syndrome, Wittmaack-Ekbom Syndrome, WM Syndrome, WMS, WNS, Wohlfart-Disease, Wohlfart-Kugelberg-Welander Disease, Wolf Syndrome, Wolf-Hirschhorn Chromosome Region (WHCR), Wolf-Hirschhorn Syndrome, Wolff-Parkinson-White Syndrome, Wolfram Syndrome, Wolman Disease (Lysosomal Acid Lypase Deficiency), Woody Guthrie's Disease, WPW Syndrome, Writer's Cramp, WS, WSS, WWS, Wyburn-Mason Syndrome, X-Linked Addison's Disease, X-linked Adrenoleukodystrophy (X-ALD), X-linked Adult Onset Spinobulbar Muscular Atrophy, X-linked Adult Spinal Muscular Atrophy, X-Linked Agammaglobulinemia with Growth Hormone Deficiency, X-Linked Agammaglobulinemia, Lymphoproliferate X-Linked Syndrome, X-linked Cardio myopathy and Neutropenia, X-Linked Centronuclear myopathy, X-linked Copper Deficiency, X-linked Copper Malabsorption, X-Linked Dominant Conradi-Hunermann Syndrome, X-Linked Dominant Inheritance Agnesis of Corpus Callosum, X-Linked Dystonia-parkinsonism, X Linked Ichthyosis, X-Linked Infantile Agammaglobulinemia, X-Linked Infantile Nectrotizing Encephalopathy, X-linked Juvenile Retinoschisis, X-linked Lissencephaly, X-linked Lymphoproliferative Syndrome, X-linked Mental Retardation-Clasped Thumb Syndrome, X-Linked Mental Retardation with Hypotonia, X-linked Mental Retardation and Macroorchidism, X-Linked Progressive Combined Variable Immunodeficiency, X-Linked Recessive Conradi-Hunermann Syndrome, X-Linked Recessive Severe Combined Immunodeficiency, X-

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Linked Retinoschisis, X-linked Spondyloepiphyseal Dysplasia, Xanthine Oxidase Deficiency (Xanthinuria Deficiency, Hereditary), Xanthinuria Deficiency, Hereditary (Xanthine Oxidase Deficiency), Xanthogranulomatosis Generalized, Xanthoma Tuberosum, Xeroderma Pigmentosum, Xeroderma Pigmentosum Dominant Type, 5 Xeroderma Pigmentosum Type A I XPA Classical Form, Xeroderma Pigmentosum Type B II XPB, Xeroderma Pigmentosum Type E V XPE, Xeroderma Pigmentosum Type C III XPC, Xeroderma Pigmentosum Type D IV XPD, Xeroderma Pigmentosum Type F VI XPF, Xeroderma Pigmentosum Type G VII XPG, Xeroderma Pigmentosum Variant Type XP-V, Xeroderma-Talipes-and Enamel Defect, Xerodermic Idiocy, Xerophthalmia, 10 Xerotic Keratitis, XLP, XO Syndrome, XP, XX Male Syndrome, Sex Reversal, XXXXX Syndrome, XXY Syndrome, XYY Syndrome, XYY Chromosome Pattern, Yellow Mutant Albinism, Yellow Nail Syndrome, YKL, Young Female Arteritis, Yunis-Varon Syndrome, YY Syndrome, Z-E Syndrome, Z- and -Protease Inhibitor Deficiency, Zellweger Syndrome, Zellweger cerebro-hepato-renal syndrome, ZES, Ziehen-Oppenheim Disease 15 (Torsion Dystonia), Zimmermann-Laband Syndrome, Zinc Deficiency Congenital, Zinsser-Cole-Engman Syndrome, ZLS, Zollinger-Ellison Syndrome.

As used herein a "cancer" refers to a group of diseases and disorders that are characterized by uncontrolled cellular growth (e.g. formation of tumor) without any differentiation of 20 those cells into specialized and different cells. Cancers which can be treated using the methods of the present invention include, without being limited to, ABL1 protooncogene, AIDS Related Cancers, Acoustic Neuroma, Acute Lymphocytic Leukaemia, Acute Myeloid Leukaemia, Adenocystic carcinoma, Adrenocortical Cancer, Agnogenic myeloid metaplasia, Alopecia, Alveolar soft-part sarcoma, Anal cancer, Angiosarcoma, Aplastic 25 Anaemia, Astrocytoma, Ataxia-telangiectasia, Basal Cell Carcinoma (Skin), Bladder Cancer, Bone Cancers, Bowel cancer, Brain Stem Glioma, Brain and CNS Tumours, Breast Cancer, CNS tumours, Carcinoid Tumours, Cervical Cancer, Childhood Brain Tumours, Childhood Cancer, Childhood Leukaemia, Childhood Soft Tissue Sarcoma, Chondrosarcoma, Choriocarcinoma, Chronic Lymphocytic Leukaemia, Chronic Myeloid 30 Leukaemia, Colorectal Cancers, Cutaneous T-Cell Lymphoma, Dermatofibrosarcoma-protuberans, Desmoplastic-Small-Round-Cell-Tumour, Ductal Carcinoma, Endocrine

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Cancers, Endometrial Cancer, Ependynoma, Esophageal Cancer, Ewing's Sarcoma, Extra-Hepatic Bile Duct Cancer, Eye Cancer, Eye: Melanoma, Retinoblastoma, Fallopian Tube cancer, Fanconi Anaemia, Fibrosarcoma, Gall Bladder Cancer, Gastric Cancer, Gastrointestinal Cancers, Gastrointestinal-Carcinoid-Tumour, Genitourinary Cancers, 5 Germ Cell Tumours, Gestational-Trophoblastic-Disease, Glioma, Gynaecological Cancers, Haematological Malignancies, Hairy Cell Leukaemia, Head and Neck Cancer, Hepatocellular Cancer, Hereditary Breast Cancer, Histiocytosis, Hodgkin's Disease, Human Papillomavirus, Hydatidiform mole, Hypercalcemia, Hypopharynx Cancer, IntraOcular Melanoma, Islet cell cancer, Kaposi's sarcoma, Kidney Cancer, Langerhan's- 10 Cell-Histiocytosis, Laryngeal Cancer, Leiomyosarcoma, Leukaemia, Li-Fraumeni Syndrome, Lip Cancer, Liposarcoma, Liver Cancer, Lung Cancer, Lymphedema, Lymphoma, Hodgkin's Lymphoma, Non-Hodgkin's Lymphoma, Male Breast Cancer, Malignant-Rhabdoid-Tumour-of-Kidney, Medulloblastoma, Melanoma, Merkel Cell Cancer, Mesothelioma, Metastatic Cancer, Mouth Cancer, Multiple Endocrine Neoplasia, 15 Mycosis Fungoides, Myelodysplastic Syndromes, Myeloma, Myeloproliferative Disorders, Nasal Cancer, Nasopharyngeal Cancer, Nephroblastoma, Neuroblastoma, Neurofibromatosis, Nijmegen Breakage Syndrome, Non-Melanoma Skin Cancer, Non-Small-Cell-Lung-Cancer-(NSCLC), Ocular Cancers, Oesophageal Cancer, Oral cavity Cancer, Oropharynx Cancer, Osteosarcoma, Ostomy Ovarian Cancer, Pancreas Cancer, 20 Paranasal Cancer, Parathyroid Cancer, Parotid Gland Cancer, Penile Cancer, Peripheral-Neuroectodermal-Tumours, Pituitary Cancer, Polycythemia vera, Prostate Cancer, Rare-cancers-and-associated-disorders, Renal Cell Carcinoma, Retinoblastoma, Rhabdomyosarcoma, Rothmund-Thomson Syndrome, Salivary Gland Cancer, Sarcoma, Schwannoma, Sezary syndrome, Skin Cancer, Small Cell Lung Cancer (SCLC), Small 25 Intestine Cancer, Soft Tissue Sarcoma, Spinal Cord Tumours, Squamous-Cell-Carcinoma-(skin), Stomach Cancer, Synovial sarcoma, Testicular Cancer, Thymus Cancer, Thyroid Cancer, Transitional-Cell-Cancer-(bladder), Transitional-Cell-Cancer-(renal-pelvis/-ureter), Trophoblastic Cancer, Urethral Cancer, Urinary System Cancer, Uroplakins, Uterine sarcoma, Uterus Cancer, Vaginal Cancer, Vulva Cancer, Waldenstrom's- 30 Macroglobulinemia, Wilms' Tumour.

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As used herein, a "brain disease or disorder" refers to any disease or disorder of the brain which results in either impaired cognitive ability or abnormal pathology. Brain diseases and disorders which can be treated using the methods of the present invention, include without being limited to, Acute Disseminated Encephalomyelitis, Arteriovenous  
5 Malformations and Other Vascular Lesions of the Central Nervous System, Cavernous Malformation, Cerebral Atrophy, Corticobasal Degeneration, Encephalopathy, Fahr's Syndrome, Kuru Moyamoya Disease, Neuronal Migration Disorders, Progressive Multifocal Leukoencephalopathy, Pseudotumor Cerebri (Benign Intracranial Hypertension), Transmissible Spongiform Encephalopathies, Wernicke-Korsakoff  
10 Syndrome, Chordoma Craniopharyngioma Medulloblastoma Meningioma Pineal Tumors Pituitary Adenoma Primitive Neuroectodermal Tumors Schwannoma Vascular Tumors, astrocytoma, glioblastomas, metastatic brain tumors, amyotrophic lateral sclerosis (ALS), progressive muscular atrophy, postpolio syndrome, Adrenoleukodystrophy, Alexander Disease, Alpers' Disease, Canavan Disease, Dementia with Lewy Bodies, Friedreich's  
15 Ataxia, Spanish Friedreich's Ataxia, Hallervorden-Spatz Disease, Krabbe Disease, Leigh's Disease, Leukodystrophy, Monomelic Amyotrophy, Olivopontocerebellar Atrophy, Opsoclonus Myoclonus, Paraneoplastic Syndromes, Pelizaeus-Merzbacher Disease, Progressive Multifocal Leukoencephalopathy, Progressive Supranuclear Palsy, Spanish Ramsay Hunt Syndrome Type II, Shy-Drager Syndrome, Alzheimer's disease,  
20 amyotrophic lateral sclerosis, aphasia, attention deficit disorder with hyperactivity, back pain, Bell's palsy, brain cancer, brain diseases, carpal tunnel syndrome, cerebral palsy, Charcot-Marie-tooth disease, Creutzfeldt-Jakob disease, degenerative nerve diseases, dementia, dizziness and vertigo, dystonia, encephalitis, epilepsy, Guillain-Barre syndrome, head and brain injuries, headache and migraine, hydrocephalus, memory, meningitis,  
25 movement disorders, multiple sclerosis, myasthenia gravis, neural tube defects, neurofibromatosis, neurologic diseases (general), pain, paralysis, Parkinson's disease, peripheral nerve disorders, phenylketonuria, pituitary disorders, reflex sympathetic dystrophy, restless legs, Reye syndrome, seizures, shingles (herpes zoster), sleep disorders, spina bifida, spinal cord diseases and injuries, spinal cord injuries, stroke, thoracic outlet  
30 syndrome, tourette syndrome, tremor, tuberous sclerosis, and West Nile virus.

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- As used herein "inflammatory diseases and disorders" encompass those disease and disorders which result in a response of redness, swelling, pain, and a feeling of heat in certain areas that is meant to protect tissues affected by injury or disease. Inflammatory diseases which can be treated using the methods of the present invention, include, without  
5 being limited to, acne, angina, arthritis, aspiration pneumonia, empyema, gastroenteritis, inflammation, intestinal flu, NEC, necrotizing enterocolitis, pelvic inflammatory disease, pharyngitis, PID, pleurisy, raw throat, redness, rubor, sore throat, stomach flu and urinary tract infections.
- 10 Examples of congenital immunodeficiency disorders of antibody production (B lymphocyte abnormalities) include hypogammaglobulinemia (lack of one or more specific antibodies), which usually causes repeated mild respiratory infections, and agammaglobulinemia (lack of all or most antibody production), which results in frequent severe infections and is often fatal. Congenital disorders affecting the T lymphocytes may  
15 cause increased susceptibility to fungi, resulting in repeated Candida (yeast) infections. Inherited combined immunodeficiency affects both T lymphocytes and B lymphocytes. It is often fatal within the first year of life because there is no resistance to disease or infection.
- 20 As used herein, the term "infertility" refers to the inability to conceive an offspring. Disease and disorders associated with an infertility which can be treated using the methods of the present invention include, without being limited to, Varicocoele, Galactorrhoea-Hyperprolactinaemia, Cryptorchism (maldescended or ectopic testis), Gonadal dysgenesis, Young's syndrome, Klinefelter's syndrome, Germinal cell aplasia, Haemochromatosis,  
25 Kallmann syndrome, Myotonic dystrophy, 5-Alpha reductase deficiency, Cystic fibrosis, Kartagener's syndrome, Incomplete androgen insensitivity, Kennedy's disease, Galactorrhoea-Hyperprolactinaemia, Hypopituitarism, Epididymo-orchitis, Pituitary tumour, Amenorrhoea (Specific type of Female infertility), Haemosiderosis, Hypokalaemic distal renal tubular acidosis, Idiopathic premature ovarian failure,  
30 Dyspareunia, Galactorrhoea-Hyperprolactinaemia, FSH receptor deficiency, Gonadal dysgenesis (female), Mullerian dystenesis, Trisomy X, Turner's syndrome, Kallmann

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syndrome, Myotonic dystrophy, C21-hydroxylase deficiency, Galactosaemia, Testicular feminization syndrome, Malabsorption syndrome, Conn's syndrome, Cushing's syndrome, Diabetes mellitus type 2, Galactorrhoea-Hyperprolactinaemia, Hyperthyroidism, Hypopituitarism, Hypothyroidism, Sheehan's syndrome, Autoimmune adrenalitis, Systemic lupus erythematosus, Adrenal cortex tumours, Pituitary tumour, Prolactin secreting pituitary tumour, Benign neoplastic conditions, Cushing's disease, Malignant neoplastic conditions, Ovarian cancer, Polycystic ovary syndrome and Pelvic inflammatory disease.

10 However, the present invention is particularly directed to identifying genotypes associated with genetic deafness or a propensity for development of genetic deafness in human subjects.

Accordingly, in a preferred embodiment, the present invention provides a method for genotyping a human with respect to a gene or target nucleic acid sequence associated with genetic deafness, said method comprising contacting an allele specific oligonucleotide immobilized to a solid support with a single-stranded form of RNA or DNA from a human to be tested labeled directly or indirectly with a reporter molecule capable of giving an identifiable signal under conditions which permit hybridization of single stranded RNA or DNA which is exactly complementary to the immobilized allele specific oligonucleotide but substantially less or no hybridization of non-complementary single-stranded RNA or DNA molecules and then screening for the presence or absence or level of reporter molecule which provides an indicator of the genetic identity of the single-stranded RNA or DNA molecule which in turn provides the genotype of the human.

25

The allele specific oligonucleotides are designed to differentially hybridize to a target nucleotide sequence based on at least one nucleotide difference. For example, a polymorphism or mutation at a single or multiple nucleotide positions may occur in genes in subjects suffering from genetic deafness or having a propensity to suffer from this disorder. An allele specific oligonucleotide is designed to either hybridize to a "mutant" form of a nucleotide sequence or to a "wild-type" form of the sequence. The term "allele-

30

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specific oligonucleotide" may also be read as "sequence-specific oligonucleotide". The term "allele" is not to impart any limitation.

The immobilized allele (i.e. sequence) specific oligonucleotides may target different  
5 polymorphisms or mutations within a single gene or may target polymorphisms or mutations in multiple genes (i.e. two or more genes). Furthermore, the allele specific oligonucleotides may cover the same or multiple mutations in two or more subjects. Consequently, the allele specific oligonucleotides are in effect an array of nucleic acid molecules which exhibit complementarity to a nucleotide sequence from a healthy subject  
10 not exhibiting genetic deafness or a nucleotide sequence from a subject exhibiting genetic deafness.

Accordingly, another aspect of the present invention contemplates a method for genotyping a subject with respect to one or a multiplicity of genes or target nucleic acid  
15 associated with genetic deafness, said method comprising contacting an array of allele specific oligonucleotides immobilized to a solid support with a single-stranded form of RNA or DNA from a subject to be tested labeled directly or indirectly with a reporter molecule capable of giving an identifiable signal wherein said single-stranded RNA or DNA comprises a nucleotide sequence identical to at least one allele specific  
20 oligonucleotide sequence or differs by at least one nucleotide from the allele specific oligonucleotide sequence, said contact being under stringency conditions which permit differential hybridization between identical nucleotide sequences and sequences having at least one mismatch and then screening for the presence, absence or level of signal from the reporter molecule wherein the pattern of presence, absence or level of signal provides the  
25 identity of the genotype of the subject.

An important key feature of the present invention is the selection of genes or target nucleic acid sequences which differ in nucleotide sequence by at least one nucleotide between a healthy subject and a subject having genetic deafness or a predisposition for development  
30 of same. Suitable genes or nucleic acid target sequences include *inter alia* *connexion 26*, *pendrin*, mitochondrial 12S rRNA and *usherin*. However, the present invention extends to

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a range of other genes or target nucleotide sequences.

Another key feature of the present invention is the selection of stringency conditions required to induce differential hybridization capacity between identically complementary  
5 nucleotide sequences and those which differ by at least one nucleotide. Useful hybridization conditions for the practice of the present invention include 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C in the following sequence:-

- 1-4 X SSC/0.05% - 0.4% SDS (1-5 min);
- 10 0.1-1 X SSC/0.05% - 0.4% SDS (2-10 min);
- 0.5 -5 X SSC (0.5-3 min);
- 2-8 X SSC/0.05% SDS (0.5-3 min); and
- 2-8 X SSC/0.05%-2% Tween (0.5-3 min).

- 15 These conditions may vary or may have to be modified for the particular genes or nucleic acid molecules being targeted. All such variations are encompassed by the present invention.

The immobilized oligonucleotides may be from about 5 to about 100 nucleotides in length  
20 although oligonucleotides outside this range are nevertheless still contemplated in accordance with the present invention. Particularly preferred oligonucleotides are from about 10 to about 50 nucleotides in length or from about 15 to about 30 nucleotides in length.

- 25 Accordingly, another aspect of the present invention provides a method for genotyping a human subject for a gene or target nucleotide sequence selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of the above genes or target nucleotide sequences is associated with genetic deafness or a propensity for genetic deafness to develop, said method comprising contacting an  
30 immobilized array of oligonucleotides which comprise a nucleotide sequence corresponding to a wild-type nucleotide sequence or a mutant nucleotide sequence or one

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or more of the above-mentioned genes or target nucleotide sequences with a single-stranded DNA molecule labeled with a reporter molecule capable of providing an identifiable signal from said human subject or group of human subjects under stringency conditions which permit differential hybridization between identical nucleotide sequences relative to nucleotide sequences which differ by at least one nucleotide and recording the presence, absence or level of signal from the reporter molecule which indicates which oligonucleotide has an identical nucleotide sequence to a DNA sequence from a human subject.

The oligonucleotides immobilized to the array are referred to herein as "allele-specific oligonucleotides". The term "allele" is not to impart any limitation as to the function of the oligonucleotides. In essence, the nucleotide sequence of the oligonucleotide will encompass one or more nucleotides in a corresponding nucleotide sequence of a gene or target nucleic acid molecule, such as from *connexin 26*, *pendrin*, mitochondrial 12S rRNA or *usherin* but where at least one nucleotide in the gene or target nucleic acid molecule may differ between a healthy subject or a subject with genetic deafness or a propensity to develop same.

The oligonucleotides may comprise nucleotide sequences at the 5' or 3' ends to facilitate less folding of the oligonucleotides or to otherwise keep the sequence specific portion further away from the solid support.

In a particular embodiment, the present invention provides a set of one or more oligonucleotides having the sequence:-

25

$$[n]_x - A$$

wherein:

30

n is one or a range of different nucleotides;

x is the length of the nucleotide sequence [n]; and

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A is a nucleotide sequence selected from SEQ ID NOs:33 to 64.

In one particular example, n is T and x is from about 5 to about 30 such as about 10. Specific examples of  $[n]_x$  - A include the oligonucleotides defined by SEQ ID NOs:1 to 32.

5 The  $[n]_x$  portion may also be a chemical linker.

In one embodiment, the oligonucleotides comprise "wild-type" nucleotide sequences meaning that the nucleotide sequences correspond to the exact same sequence as in a gene or target from a healthy subject. In this case, if a single-stranded DNA sequence from one  
10 of the aforementioned genes or nucleic acid targets differs by at least one nucleotide from the oligonucleotide sequence, then, under the differential hybridization conditions employed, a DNA with non-identical nucleotide sequence will not substantially hybridize.

Similarly, the oligonucleotides could encompass nucleotide sequences which are derived  
15 from a mutated gene. In this case, only DNA from subjects with a mutated gene or target nucleic acid would substantially hybridize. The present invention encompasses both forms of arrays.

Accordingly, another aspect of the present invention contemplates a method for  
20 genotyping a human subject from a gene or nucleic acid target selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label directly or indirectly into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence  
25 corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:64 under stringency conditions such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for  
30 hybridization by measuring a signal or level of signal from the label.

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The nucleotide sequence of the target nucleotide sequence may be modified up- or down-stream of a mutation to be detected or groups of mutations to be detected. This may be useful to interrupt a particular sequence of nucleotides to improve hybridization sensitivity. For example, mismatched primers may be used to introduce a mismatch within a sequence  
5 of G residues. This may be useful, for example, in relation to the target DNA sequence which hybridizes to a 35ΔG mutation in *connexion 26*. SEQ ID NOs:1 to 4, for example, include a sequence of six Gs. This sequence is disruptable by a non-G nucleotide. This is proposed to reduce oligonucleotide bending and improve hybridization efficiency or sensitivity.

10

This approach applies to all the oligonucleotides of the present invention.

Preferably, the stringency conditions comprise 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C in the following sequence:-

15

1-4 X SSC/0.05% - 0.4% SDS (1-5 min);  
0.1-2 X SSC/0.05% - 0.4% SDS (2-10 min);  
0.5 X -5 X SSC (0.5-3 min);  
2-8 X SSC/0.05% (0.5-3 min); and  
20 2-8 X SSC/0.05%-2% Tween (0.5-3 min).

20

Consequently, another aspect of the present invention is directed to a method for genotyping a human subject from a gene or nucleic acid target selected from *connexion 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these  
25 genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide  
30 selected from SEQ ID NO:1 to SEQ ID NO:32 under stringency conditions of 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C in the following

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sequence:-

1-4 X SSC/0.05% - 0.4% SDS (1-5 min);

0.1-3 X SSC/0.05% - 0.4% SDS (2-10 min);

5 0.5 X -5 X SSC (0.5-3 min);

2-8 X SSC/0.05% (0.5-3 min); and

2-8 X SSC/0.05%-2% Tween (0.5-3 min);

such that substantially only identically complementary DNA from the subject is capable of  
10 hybridizing to the corresponding immobilized oligonucleotide and screening for  
hybridization by measuring a signal or level of signal from the label.

Any of a number of labels may be incorporated into the amplified test DNA. Fluorescent  
labels and fluorophores are particularly useful.

15

In one embodiment, a few cycles (e.g. 1 or 2 or 3 or 4 or 5) PCR is conducted using pairs  
of primers, one or both of which are generally labeled with the same or a different reporter  
molecule capable of giving a distinguishable signal. The use of fluorophores is particularly  
useful in the practice of the present invention. Examples of suitable fluorophores may be  
20 selected from the list given in Table 2. Other labels include luminescence and  
phosphorescence as well as infrared dyes. These dyes or fluorophores may also be used as  
reporter molecules for antibodies.

**TABLE 2** List of suitable fluorophores

25

Probe	Ex <sup>1</sup> (nm)	Em <sup>2</sup> (nm)
Hydroxycoumarin	325	386
Aminocoumarin	350	455
Methoxycoumarin	360	410
Cascade Blue	375; 400	423
Lucifer Yellow	425	528
NBD	466	539
R-Phycoerythrin (PE)	480; 565	578
PE-Cy5 conjugates	480; 565; 650	670

Probe	Ex <sup>1</sup> (nm)	Em <sup>2</sup> (nm)
PE-Cy7 conjugates	480; 565; 743	767
APC-Cy7 conjugates	650; 755	767
Red 613	480; 565	613
Fluorescein	495	519
FluorX	494	520
BODIPY-FL	503	512
TRITC	547	574
X-Rhodamine	570	576
Lissamine Rhodamine B	570	590
PerCP	490	675
Texas Red	589	615
Allophycocyanin (APC)	650	660
TruRed	490, 675	695
Alexa Fluor 350	346	445
Alexa Fluor 430	430	545
Alexa Fluor 488	494	517
Alexa Fluor 532	530	555
Alexa Fluor 546	556	573
Alexa Fluor 555	556	573
Alexa Fluor 568	578	603
Alexa Fluor 594	590	617
Alexa Fluor 633	621	639
Alexa Fluor 647	650	688
Alexa Fluor 660	663	690
Alexa Fluor 680	679	702
Alexa Fluor 700	696	719
Alexa Fluor 750	752	779
Cy2	489	506
Cy3	(512); 550	570; (615)
Cy3,5	581	596; (640)
Cy5	(625); 650	670
Cy5,5	675	694
Cy7	743	767
Hoeschst 33342	343	483
DAPI	345	455
Hoechst 33258	345	478
SYTOX Blue	431	480
Chromomycin A3	445	575
Mithramycin	445	575
YOYO-1	491	509
SYTOX Green	504	523
SYTOX Orange	547	570
Ethidium Bromide	493	620
7-AAD	546	647

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Probe	Ex <sup>1</sup> (nm)	Em <sup>2</sup> (nm)
Acridine Orange	503	530/640
TOTO-1, TO-PRO-1	509	533
Thiazole Orange	510	530
Propidium Iodide (PI)	536	617
TOTO-3, TO-PRO-3	642	661
LDS 751	543; 590	712; 607
Y66F	360	508
Y66H	360	442
EBFP	380	440
Wild-type	396, 475	50, 503
GFPuv	385	508
ECFP	434	477
Y66W	436	485
S65A	471	504
S65C	479	507
S65L	484	510
S65T	488	511
EGFP	489	508
EYFP	514	527
DsRed	558	583
Monochlorobimane	380	461
Calcein	496	517

<sup>1</sup> Ex: Peak excitation wavelength (nm)

<sup>2</sup> Em: Peak emission wavelength (nm)

5 Any suitable method of analyzing fluorescence emission is encompassed by the present invention. In this regard, the invention contemplates techniques including but not restricted to 2-photon and 3-photon time resolved fluorescence spectroscopy as, for example, disclosed by Lakowicz *et al.*, *Biophys. J.* 72: 567, 1997, fluorescence lifetime imaging as, for example, disclosed by Eriksson *et al.*, *Biophys. J.* 2: 64, 1993 and fluorescence  
 10 resonance energy transfer as, for example, disclosed by Youvan *et al.*, *Biotechnology et alia* 3: 1-18, 1997.

Luminescence and phosphorescence may result respectively from a suitable luminescent or phosphorescent label as is known in the art. Any optical means of identifying such label  
 15 may be used in this regard.

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Infrared radiation may result from a suitable infrared dye. Exemplary infrared dyes that may be employed in the invention include but are not limited to those disclosed in Lewis *et al.*, *Dyes Pigm.* 42(2): 197, 1999, Tawa *et al.*, *Mater. Res. Soc. Symp. Proc.* 488  
5 [Electrical, Optical and Magnetic Properties of Organic Solid-State Materials IV], 885-890, Daneshvar *et al.*, *J. Immunol. Methods* 226(1-2): 119-128, 1999, Rapaport *et al.*, *Appl. Phys. Lett.* 74(3): 329-331, 1999 and Durig *et al.*, *J. Raman Spectrosc.* 24(5): 281-285, 1993. Any suitable infrared spectroscopic method may be employed to interrogate the infrared dye. For instance, fourier transform infrared spectroscopy as, for example,  
10 described by Rahman *et al.*, *J. Org. Chem.* 63: 6196, 1998 may be used in this regard.

Suitably, electromagnetic scattering may result from diffraction, reflection, polarization or refraction of the incident electromagnetic radiation including light and X-rays. Such scattering can be used to quantitate the level of mRNA or level of protein.

15

Flow cytometry is particularly useful in analyzing fluorophore emission.

As is known in the art, flow cytometry is a high throughput technique which involves rapidly analyzing the physical and chemical characteristics of particles (e.g. labeled DNA)  
20 as they pass through the path of one or more laser beams while suspended in a fluid stream. As each particle intercepts the laser beam, the scattered light and fluorescent light emitted by each cell or particle is detected and recorded using any suitable tracking algorithm as, for example, described hereunder.

25 A modern flow cytometer is able to perform these tasks up to 100,000 cells/particles  $s^{-1}$ . Through the use of an optical array of filters and dichroic mirrors, different wavelengths of fluorescent light can be separated and simultaneously detected. In addition, a number of lasers with different excitation wavelengths may be used. Hence, a variety of fluorophores can be used to target and examine, for example, different immune effectors within a  
30 sample or immune effectors from multiple subjects.

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Suitable flow cytometers which may be used in the methods of the present invention include those which measure five to nine optical parameters (see Table 3) using a single excitation laser, commonly an argon ion air-cooled laser operating at 15 mW on its 488 nm spectral line. More advanced flow cytometers are capable of using multiple excitation  
 5 lasers such as a HeNe laser (633 nm) or a HeCd laser (325 nm) in addition to the argon ion laser (488 or 514 nm).

**TABLE 3** Exemplary optical parameters which may be measured by a flow cytometer.

Parameter	Acronym	Detection angle from incident laser beam	Wavelength (nm)
Forward scattered light	FS	2-5°	488
Side scattered light	SS	90°	488
"Green" fluorescence	FL1	90°	510-540 <sup>†</sup>
"Yellow" fluorescence	FL2	90°	560-580 <sup>†</sup>
"Red" fluorescence	FL3	90°	>650 <sup>#</sup>

10

\* using a 488 nm excitation laser

† width of bandpass filter

# longpass filter

15 For example, Biggs *et al.*, *Cytometry* 36: 36-45, 1999 have constructed an 11-parameter flow cytometer using three excitation lasers and have demonstrated the use of nine distinguishable fluorophores in addition to forward and side scatter measurements for purposes of immunophenotyping (i.e. classifying) particles. The maximum number of parameters commercially available currently is 17: forward scatter, side scatter and three  
 20 excitation lasers each with five fluorescence detectors. Whether all of the parameters can be adequately used depends heavily on the extinction coefficients, quantum yields and amount of spectral overlap between all fluorophores (Malemed *et al.*, "Flow cytometry and sorting", 2<sup>nd</sup> Ed., New York, Wiley-Liss, 1990). However, it will be understood that the present invention is not restricted to any particular flow cytometer or any particular set of  
 25 parameters. In this regard, the invention also contemplates use in place of a conventional flow cytometer, a microfabricated flow cytometer as, for example, disclosed by Fu *et al.*,

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*Nature Biotechnology* 17: 1109-1111, 1999.

Electrodes may also be used as a detection system. Such a system relies on complementary binding of RNA or DNA to assemble an electronic circuit which thereby  
5 creates a detectable electronic signal. One particularly useful system is eSensor (trade mark; Motorola) which is well described at [http://www.motorola.com/lifesciences/esensor/tech\\_overview.html](http://www.motorola.com/lifesciences/esensor/tech_overview.html).

The signal produced following hybridization provides a genotype index (GI).

10

The GI is calculated by the algorithm:-

$$GI = \frac{SV_N}{SV_N + SV_M}$$

15 wherein:

$SV_N$  is the normal spot value; and

$SV_M$  is the mutant spot value.

20 Generally, a background subtracted median pixel intensity is used as the spot value.

Accordingly, a method for genotyping a human subject from a gene or nucleic acid target selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a  
25 propensity to develop genetic deafness, said method incorporating a label into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:32 under stringency  
30 conditions of 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C

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in the following sequence:-

- 1-4 X SSC/0.05% - 0.4% SDS (1-5 min);
- 0.1-4 X SSC/0.05% - 0.4% SDS (2-10 min);
- 5 0.5 X -5 X SSC (0.5-3 min);
- 2-8 X SSC/0.05% (0.5-3 min); and
- 2-8 X SSC/0.05%-2% Tween (0.5-3 min);

such that substantially only identically complementary DNA from the subject is capable of  
 10 hybridizing to the corresponding immobilized oligonucleotide and screening for  
 hybridization by measuring a signal or level of signal from the label, wherein a GI value is  
 determined by the algorithm:-

$$GI = \frac{SV_N}{SV_N + SV_M}$$

15

wherein:

$SV_N$  is the normal spot value; and

$SV_M$  is the mutant spot value;

20

such that:

if  $0.8 < GI < 1.0$ , then the genotype is N/N;

if  $0.65 < GI < 0.5$ , then the genotype is N/M; and

25

if  $0.0 < GI < 0.2$ , then the genotype is M/M;

wherein:

N is a normal allele; and

30

M is a mutant allele.

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The present invention further contemplates an array of oligonucleotides selected from two or more of SEQ ID NOs:1 to 32 for use in a differential hybridization assay of DNA from a subject being tested for genetic deafness or a propensity for development of genetic  
5 deafness.

This aspect of the present invention provides a kit for use in screening subjects for the presence of genes or nucleic acid molecules such as mitochondrial 12S rRNA which are either "mutated" or "normal" (i.e. wild-type). A mutant gene or target is proposed to be  
10 associated with genetic deafness or a predisposition for developing genetic deafness. A "normal" gene or target is from a subject without genetic deafness.

The present method is also useful in designing therapeutic protocols for treating genetic deafness. A therapeutic protocol includes medical intervention as well as behavioral  
15 changes required by a subject who is likely to become deaf.

The present invention is further described by the following non-limiting Examples.

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**EXAMPLE 1*****DNA preparation***

1. Amplify patient DNA in three PCR reactions containing primer mixes 1, 2 and 3  
 5 (Table 35) according to Table 4:

**TABLE 4** PCR reaction mixtures

VOLUME	CONTENT
$x \mu\text{l}^1$	(50 ng) patient DNA
2.5 $\mu\text{l}$	10X Taq buffer
2.5 $\mu\text{l}$	10X primer mix
2.5 $\mu\text{l}$	10X nucleotide labeling mix
0.5 $\mu\text{l}$	Taq polymerase
$y \mu\text{l}^2$	dH <sub>2</sub> O
25 $\mu\text{l}$	

10 <sup>1</sup> volume required to provide 50 ng of DNA

<sup>2</sup> volume required to make up to 25  $\mu\text{l}$

PCR is one cycle of denaturation for 5 min at 94° followed by 40 cycles of  
 denaturation for 30 s at 94°C, annealing for 30 s at 58°C and extension for 30 s at  
 15 72°C, followed by a final extension step for 5 min at 72°C.

2. Take 5  $\mu\text{l}$  of each reaction for gel analysis (optional), pool the remaining DNA into  
 one tube and purify on a Qiagen MinElute column according to the manufacturer's  
 instructions. Elute in 12  $\mu\text{l}$  10 mM Tris-Cl pH 7.5.

20

3. Add 3  $\mu\text{l}$  5X T7 gene 6 exonuclease buffer and 0.5  $\mu\text{l}$  T7 gene 6 exonuclease.  
 Incubate 20 min at 37°C, then heat inactivate at 90°C for 10 min.

4. Touch spin to collect condensate and store at -20°C until use.

**TABLE 5** Primer mixers<sup>1</sup>

5

<b>Primer Mix 1</b>	286 bp, 137 bp
P1	TCTTTTCCAGAGCAAACCGC [SEQ ID NO:65]
P3-thio	GsAsCsAsCsGAAGATCAGCTGCAG [SEQ ID NO:66]
h.48	CGTCACCCTCCTCAAGTATACTTC [SEQ ID NO:67]
h.6-thio	GsCsTsTsTsGTGTAAAGCTACACTCTGG [SEQ ID NO:68]
<b>Primer Mix 2</b>	248 bp, 217 bp, 139 bp
PDS6F	GGTTTCTATCTCAGGCAAACAT [SEQ ID NO:69]
PDS6R-thio	AsTsTsGsTsTTCTGGAATGAACAGTGACC [SEQ ID NO:70]
PDS8F2	TTCAGACGATAATTGCTACTG [SEQ ID NO:71]
PDS8R-thio	GsAsCsTsGsACTTACTGACTTAATG [SEQ ID NO:72]
PDS10F	GTAGGATCGTTGTCATCCAG [SEQ ID NO:73]
PDS10R-thio	CsGsAsGsCsCTTCCTCTGTTGC [SEQ ID NO:74]
<b>Primer Mix 3</b>	311 bp, 159 bp
P4	CTGCAGCTGATCTTCGTGTC [SEQ ID NO:75]
P7-thio	AsCsAsAsAsGCAGTCCACAGTGTT [SEQ ID NO:76]
USH2AF	ATGTGAGCCCTGCCAGTGTA [SEQ ID NO:77]
USH2AR-thio	TsCsAsCsAsGGCCTTACAATTGGTG [SEQ ID NO:78]

s phosphorothioate bond

<sup>1</sup> 10X primer mixes are 4  $\mu$ M each primer.

# 10 Notes

- 10X nucleotide labeling mix = 2 mM dATP, dCTP, dGTP, 1.5 mM dTTP and 0.5 mM biotin-dUTP.
- A hot start PCR protocol is preferred.

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- T7 gene 6 exonuclease can be obtained from USB (Cat. No. 70025). It is used to prepare ss DNA by selectively digesting the strand that is not protected by a phosphorothioated primer.

5

**EXAMPLE 2*****Hybridization and labeling***

1. Add 5 µl pooled ss PCR products to 5 µl hybridization buffer. Mix thoroughly.
- 10 2. Denature 5 min at 90°C.
3. Snap cool hybridization mix on ice.
4. Touch spin to collect condensate and pipette 10 µl hybridization mix onto a clean  
15 coverslip. Lower the measuring region of the chip onto the coverslip and let the hybridization mix spread to the edges of the coverslip.
5. Put the clip into a hybridization cassette containing 2X SSC in the humidification wells and incubate in a 45°C water bath for 30 min.
- 20 6. Wash chip at 45°C in the following sequence:

2X SSC/0.1% w/v SDS	3 min
0.5X SSC/0.1% w/v SDS	5 min
25 2X SSC	1 min
4X SSC/0.2% w/v Tween	1 min.
7. Let chip drain briefly but *do not allow to dry out*. Pipette 12 µl streptavidin-Cy5 diluted 1:250 in blocking solution onto a coverslip, avoiding bubbles. Lower the  
30 measuring region of the chip onto the coverslip and let the solution spread to the edges of the coverslip. Incubate in a damp chamber in the dark at RT for 30-60

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min.

8. Wash the chip 2 X 3 min in 4X SSC/0.2% w/v Tween at 45°C.
- 5 9. Rinse the chip in 0.1X SSC at RT for 2 min.
10. Dry chip by centrifugation in a 50 mL Falcon tube at 500 rpm for 3 min and store in dark, dry place until scanning.

10 Notes

- Hybridization buffer = 5X SSPE/0.01% v/v Triton X100.
  - Blocking solution = 4X SSC/0.2% w/v Tween20/5% w/v BSA.
  - Streptavidin-Cy5 can be obtained from a number of suppliers (e.g. Amersham PA45001).
- 15

**EXAMPLE 3**

*Scanning and analysis*

- 20 1. Scan the chip in a standard microarray scanner using the red Cy5 channel (635 nm).
2. Quantitate spot intensities using the scanner software. At this time, visually inspect the array and exclude any "bad" spots (e.g. poor printing or hybridization, contamination by dust particle, etc.).
- 25 3. Import results into Excel. Using the background, subtract median pixel intensity as your Spot Value (SV), calculate the Genotype Index (GI) for each normal and mutant spot pair.

30

$$GI = SV_N / (SV_N + SV_M)$$

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where  $SV_N$  = normal Spot Value and  $SV_M$  = mutant Spot Value

4. Average GI values for replicate spot pairs and use to call genotype for each mutation.

#### EXAMPLE 4

##### *Attachment of oligonucleotides*

Oligonucleotides are attached to the solid support by coupling via an epoxide group on the solid support. This is shown in Figure 1.

#### EXAMPLE 5

##### *Microarray based genotyping*

Figure 2 shows the principle of microarray genotyping. Oligonucleotides covering mutant or normal sequences are immobilized to a solid support using the coupling reaction described in Example 4. A single-stranded labeled DNA from a test substrate is then brought into contact using hybridization conditions which facilitate differential hybridization. A signal is then measured to ascertain binding or no binding.

#### EXAMPLE 6

##### *Genotyping of connexin 26 35ΔG and M34T mutations*

Figure 3 shows the results of the microarray assay. The genotypes N/N, 35ΔG/M34T, 35ΔG/35ΔG and M34T/M34T are clearly discernible.

The intensity of the signal provides a means of calculating the GI.

The GI is calculated as follows:-

$$GI = \frac{SV_N}{SV_N + SV_M}$$

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Figure 4 shows a graphical representation of the GI for the connexin 26 35ΔG and M34T mutations.

- 5 This experiment is repeated using a greater range of oligonucleotides. The results are shown in Figure 5. A genotypic graph is shown in Figure 7.

#### EXAMPLE 7

##### *Genotyping pendrin and 12S rRNA mutations*

10

Figure 6 shows genotyping of *pendrin* and 12S rRNA mutations.

A genotypic summary is shown in Figure 7.

- 15 Figure 8 summarizes the results of applying the GI to deciding whether a subject is normal (N) homozygous, N heterozygous or a mutant (M) homozygous.

#### EXAMPLE 8

##### *Potential interactions between deafness genes*

20

Figure 9 shows the results of a potential interaction between deafness genes.

- Those skilled in the art will appreciate that the invention described herein is susceptible to variations and modifications other than those specifically described. It is to be understood  
25 that the invention includes all such variations and modifications. The invention also includes all of the steps, features, compositions and compounds referred to or indicated in this specification, individually or collectively, and any and all combinations of any two or more of said steps or features.

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## CLAIMS:

1. A method for genotyping a subject with respect to a gene or target nucleic acid sequence associated with a pathological condition, said method comprising contacting an allele specific oligonucleotide immobilized to a solid support with a single-stranded form of RNA or DNA from a subject to be tested labeled directly or indirectly with a reporter molecule capable of giving an identifiable signal under conditions which permit hybridization of single stranded RNA or DNA which is exactly complementary to the immobilized allele specific oligonucleotide but substantially less or no hybridization of non-complementary single-stranded RNA or DNA molecules and then screening for the presence or absence or level of reporter molecule which provides an indicator of the genetic identity of the single-stranded RNA or DNA molecule which in turn provides the genotype of the subject.
2. The method of Claim 1 wherein the RNA or DNA from the test subject is directly labeled with labeled nucleotides incorporated *via* polymer chain reaction (PCR).
3. The method of Claim 1 wherein the RNA or DNA from the test subject is indirectly labeled with labeled nucleotides *via* hybridization of a labeled oligonucleotide to the test RNA or DNA.
4. The method of Claim 1 wherein the subject is selected for a human, a non-human primate, a livestock animal, a laboratory test animal, a companion animal and a captured wild animal.
5. The method of Claim 1 wherein the subject is a human.
6. The method of Claim 5 wherein the pathological condition is selected from an autoimmune disease, inflammatory condition, cancer, neurological disorder and a neurodegenerative disorder.

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7. The method of claim 1 wherein the pathological condition is genetic deafness or a propensity for development of genetic deafness.
8. The method of Claim 1 wherein the pathological condition is associated with genetic deafness.
9. The method of Claim 1 wherein the hybridization step is under differential hybridization conditions which permits differential hybridization between identical nucleotide sequences and sequences having at least one mismatch and the identity of the genotype of the subject is determined by the presence, absence or level of signal from the reporter molecule.
10. The method of Claim 1 or 7 or 8 wherein the RNA or DNA is *connexin 26*, *pendrin*, mitochondrial 12S rRNA or *usherin*.
11. The method of Claim 1 wherein the hybridization conditions comprise hybridization in the presence of 1-4 X SSC at 30-50°C for 15-90 min followed by washing at 30-50°C in the following sequence:
  - 1-4 X SSC/0.05% - 0.4% SDS (1-5 min);
  - 0.1-1 X SSC/0.05% - 0.4% SDS (2-10 min);
  - 0.5 X - 5 X SSC (0.5-3 min);
  - 2-8 X SSC/0.05% (0.5-3 min); and
  - 2-8 X SSC/0.05%-2% Tween (0.5-3 min).
12. The method of Claim 1 wherein the immobilized oligonucleotides are from about 5 to about 100 nucleotides in length.
13. The method of Claim 12 wherein the immobilized oligonucleotides are from about 10 to about 30 nucleotides in length.
14. The method of Claim 12 wherein the immobilized oligonucleotides are from about

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15 to about 30 nucleotides in length.

15. The method of Claim 12 wherein the immobilized oligonucleotides are selected from SEQ ID NO:1 to 64.

16. The method of claim 1 wherein a sequence of nucleotides is interrupted up- or down-stream of the immobilized oligonucleotide to improve hybridization sensitivity.

17. The method of Claim 16 wherein the interruption is in a sequence of G residues.

18. A method for genotyping a human subject from a gene or nucleic acid target selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label directly or indirectly into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:64 under stringency conditions such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for hybridization by measuring a signal or level of signal from the label.

19. A method for genotyping a human subject from a gene or nucleic acid target selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:32 under stringency conditions of 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C

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in the following sequence:-

1-4 X SSC/0.05% - 0.4% SDS (1-5 min);  
0.1-5 X SSC/0.05% - 0.4% SDS (2-10 min);  
0.5 X -5 X SSC (0.5-3 min);  
2-8 X SSC/0.05% (0.5-3 min); and  
2-8 X SSC/0.05%-2% Tween (0.5-3 min);

such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for hybridization by measuring a signal or level of signal from the label.

20. A method for genotyping a human subject from a gene or nucleic acid target selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:32 under stringency conditions of 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C in the following sequence:-

1-4 X SSC/0.05% - 0.4% SDS (1-5 min);  
0.1-5 X SSC/0.05% - 0.4% SDS (2-10 min);  
0.5 X -5 X SSC (0.5-3 min);  
2-8 X SSC/0.05% (0.5-3 min); and  
2-8 X SSC/0.05%-2% Tween (0.5-3 min);

such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for

- 100 -

hybridization by measuring a signal or level of signal from the label, wherein a GI value is determined by the algorithm:-

$$GI = \frac{SV_N}{SV_N + SV_M}$$

wherein:

$SV_N$  is the normal spot value; and

$SV_M$  is the mutant spot value;

such that:

if  $0.8 < GI < 1.0$ , then the genotype is N/N;

if  $0.65 < GI < 0.5$ , then the genotype is N/M; and

if  $0.0 < GI < 0.2$ , then the genotype is M/M;

wherein:

N is a normal allele; and

M is a mutant allele.

21. A set of one or more oligonucleotides having the sequence:-

$$[n]_x \sim A$$

wherein:

n is one or a range of different nucleotides;

x is the length of the nucleotide sequence [n]; and

A is a nucleotide sequence selected from SEQ ID NOs:33 to 64.

22. The set of one or more oligonucleotides of claim 21 wherein n is T.

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23. The set of one or more oligonucleotides of claim 21 or 22 wherein x is from about 5 to about 30.

24. The set of one or more nucleotides of claim 21 wherein  $[n]_x - A$  is selected from SEQ ID Nos: 1 to 32.

25. A kit comprising one or more oligonucleotides of any one of claims 21 to 24.

# CLAIMS:

1. A method for genotyping a subject with respect to a gene or target nucleic acid sequence selected from connexin 26, pendrin, mitochondrial 12S rRNA or ussherlin associated with a pathological condition, said method comprising contacting an allele specific oligonucleotide immobilized to a solid support with a single-stranded form of RNA or DNA from a subject to be tested labeled directly or indirectly with a reporter molecule capable of giving an identifiable signal under conditions which comprise hybridization in the presence of 1-4 X SSC at 30-50°C for 15-90 min followed by washing at 30-50°C in the following sequence:

- 1-4 X SSC/0.05% - 0.4% SDS (1-5 min);
- 0.1-1 X SSC/0.05% - 0.4% SDS (2-10 min);
- 0.5 X -5 X SSC (0.5-3 min);
- 2-8 X SSC/0.05% (0.5-3 min); and
- 2-8 X SSC/0.05%-2% Tween (0.5-3 min).

which permit hybridization of single stranded RNA or DNA which is exactly complementary to the immobilized allele specific oligonucleotide but substantially less or no hybridization of non-complementary single-stranded RNA or DNA molecules and then screening for the presence or absence or level of reporter molecule which provides an indicator of the genetic identity of the single-stranded RNA or DNA molecule which in turn provides the genotype of the subject.

2. The method of Claim 1 wherein the RNA or DNA from the test subject is directly labeled with labeled nucleotides incorporated *via* polymer chain reaction (PCR).

3. The method of Claim 1 wherein the RNA or DNA from the test subject is indirectly labeled with labeled nucleotides *via* hybridization of a labeled oligonucleotide to the test RNA or DNA.

4. The method of Claim 1 wherein the subject is selected for a human, a non-human primate, a livestock animal, a laboratory test animal, a companion animal and a captured

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wild animal.

5. The method of Claim 1 wherein the subject is a human.
6. The method of Claim 5 wherein the pathological condition is selected from an autoimmune disease, inflammatory condition, cancer, neurological disorder and a neurodegenerative disorder.
7. The method of claim 1 wherein the pathological condition is genetic deafness or a propensity for development of genetic deafness.
8. The method of Claim 1 wherein the pathological condition is associated with genetic deafness.
9. The method of Claim 1 wherein the hybridization step is under differential hybridization conditions which permits differential hybridization between identical nucleotide sequences and sequences having at least one mismatch and the identity of the genotype of the subject is determined by the presence, absence or level of signal from the reporter molecule.
10. The method of Claim 1 wherein the immobilized oligonucleotides are from about 5 to about 100 nucleotides in length.
11. The method of Claim 12 wherein the immobilized oligonucleotides are from about 10 to about 30 nucleotides in length.
12. The method of Claim 12 wherein the immobilized oligonucleotides are from about 15 to about 30 nucleotides in length.
13. The method of Claim 12 wherein the immobilized oligonucleotides are selected from SEQ ID NO:1 to 64.

- 98 -

14. The method of claim 1 wherein a sequence of nucleotides is interrupted up- or down-stream of the immobilized oligonucleotide to improve hybridization sensitivity.

15. The method of Claim 16 wherein the interruption is in a sequence of G residues.

16. A method for genotyping a human subject from a gene or nucleic acid target selected from *connexion 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label directly or indirectly into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:64 under stringency conditions such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for hybridization by measuring a signal or level of signal from the label.

17. A method for genotyping a human subject from a gene or nucleic acid target selected from *connexion 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:32 under stringency conditions of 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C in the following sequence:-

1-4 X SSC/0.05% - 0.4% SDS (1-5 min);

0.1-5 X SSC/0.05% - 0.4% SDS (2-10 min);

- 99 -

0.5 X -5 X SSC (0.5-3 min);  
2-8 X SSC/0.05% (0.5-3 min); and  
2-8 X SSC/0.05%-2% Tween (0.5-3 min);

such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for hybridization by measuring a signal or level of signal from the label.

20. A method for genotyping a human subject from a gene or nucleic acid target selected from *connexin 26*, *pendrin*, mitochondrial 12S rRNA and *usherin* wherein a mutation in one or more of these genes or targets is indicative of genetic deafness or a propensity to develop genetic deafness, said method incorporating a label into genomic DNA amplified from the human subject to be tested using primers which flank a DNA sequence corresponding to a potential mutation in a gene or nucleic acid target listed above and contacting single-stranded labeled forms of the amplified DNA with an immobilized oligonucleotide selected from SEQ ID NO:1 to SEQ ID NO:32 under stringency conditions of 1-4 X SSC at 30-50°C for 15 min to 90 min followed by washing at 30-50°C in the following sequence:-

1-4 X SSC/0.05% - 0.4% SDS (1-5 min);  
0.1-6 X SSC/0.05% - 0.4% SDS (2-10 min);  
0.5 X -5 X SSC (0.5-3 min);  
2-8 X SSC/0.05% (0.5-3 min); and  
2-8 X SSC/0.05%-2% Tween (0.5-3 min);

such that substantially only identically complementary DNA from the subject is capable of hybridizing to the corresponding immobilized oligonucleotide and screening for hybridization by measuring a signal or level of signal from the label, wherein a GI value is determined by the algorithm:-

- 100 -

$$GI = \frac{SV_N}{SV_N + SV_M}$$

wherein:

$SV_N$  is the normal spot value; and

$SV_M$  is the mutant spot value;

such that:

if  $0.8 < GI < 1.0$ , then the genotype is N/N;

if  $0.65 < GI < 0.5$ , then the genotype is N/M; and

if  $0.0 < GI < 0.2$ , then the genotype is M/M;

wherein:

N is a normal allele; and

M is a mutant allele.

21. A set of one or more oligonucleotides having the sequence:-

$$[n]_1 = A$$

wherein:

n is one or a range of different nucleotides;

$x$  is the length of the nucleotide sequence  $[n]$ ; and

A is a nucleotide sequence selected from SEQ ID NOs:33 to 64.

22. The set of one or more oligonucleotides of claim 21 wherein n is T.

23. The set of one or more oligonucleotides of claim 21 or 22 wherein x is from about 5 to about 30.



## Step 2: Genotyping of pendrin and 12S rRNA mutations

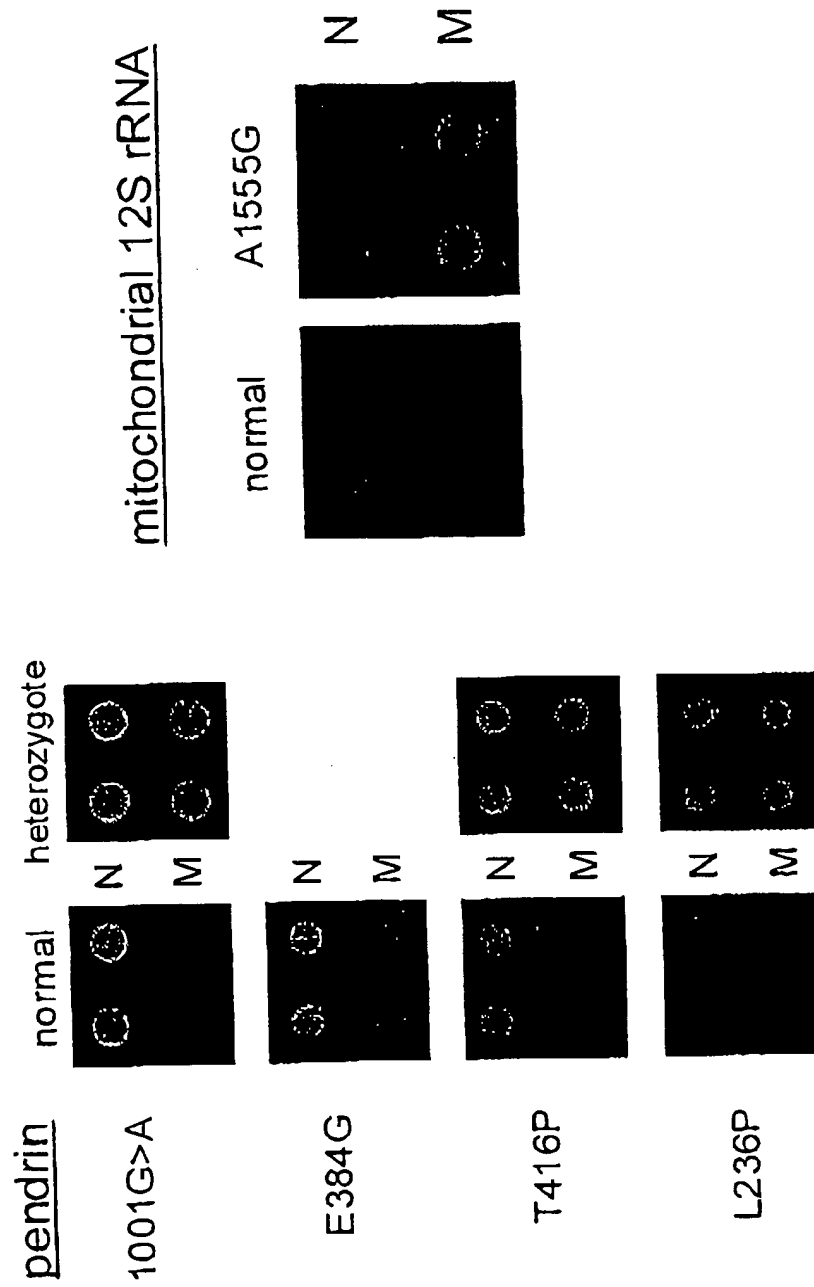


Figure 6

# Genotyping summary

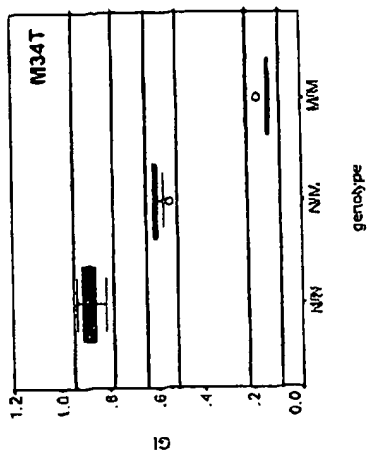


Figure 7A

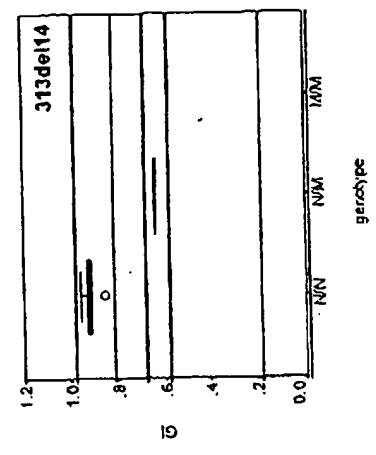


Figure 7B

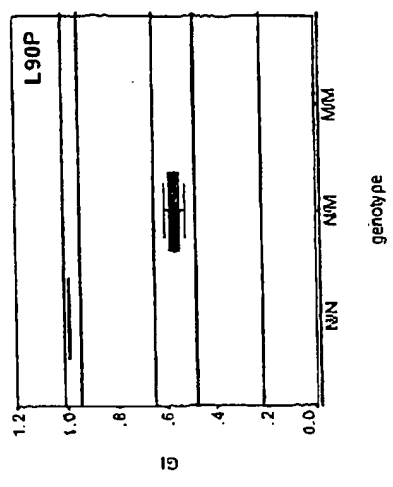


Figure 7C

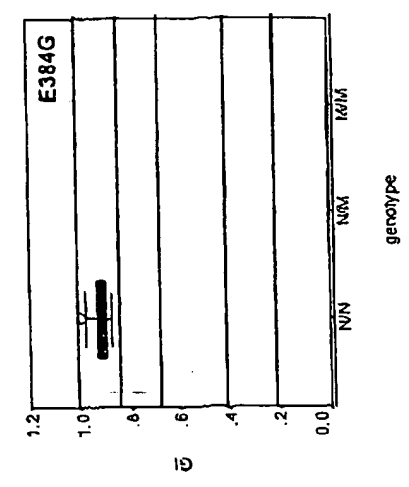


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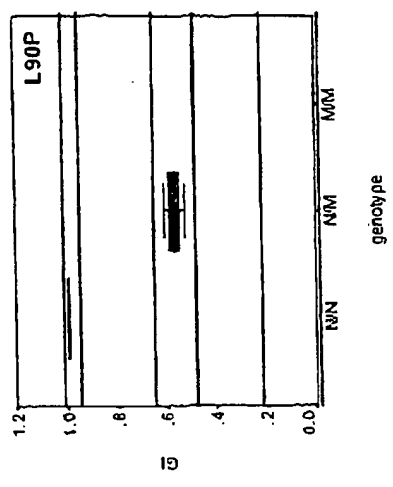


Figure 7E

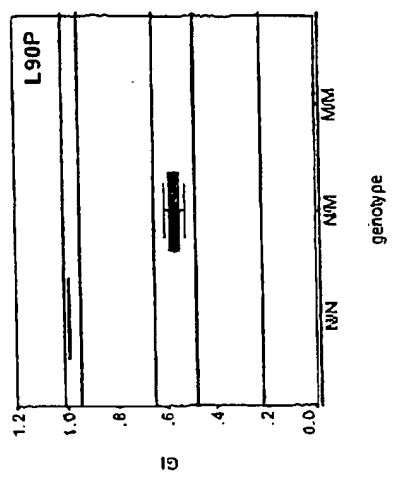


Figure 7F

# Genotyping summary

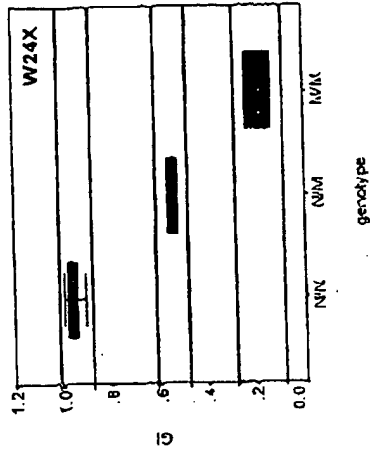


Figure 7K

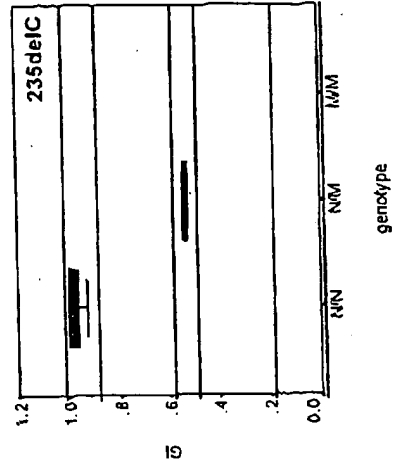


Figure 7L

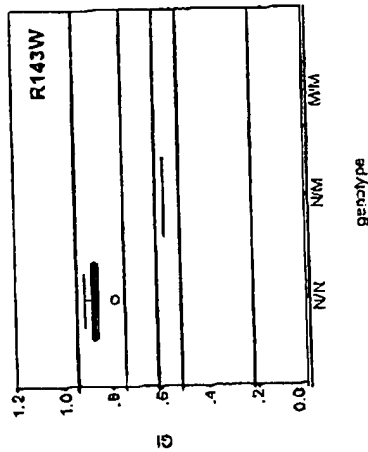


Figure 7I

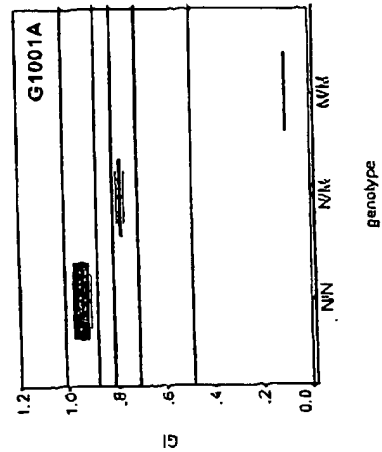


Figure 7J

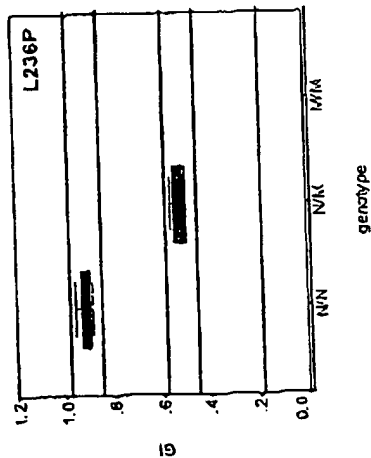


Figure 7G

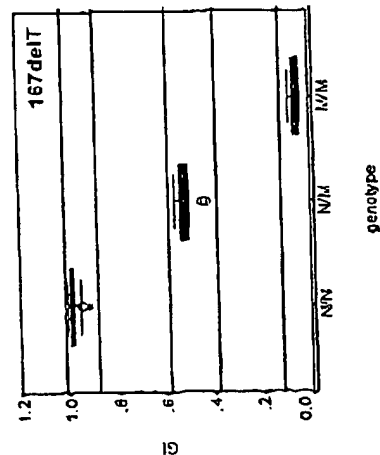


Figure 7H

WO 2004/046388

9/11

PCT/AU2003/001544

# Genotyping summary

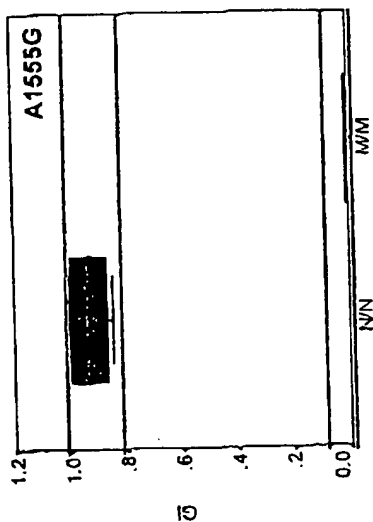


Figure 7M

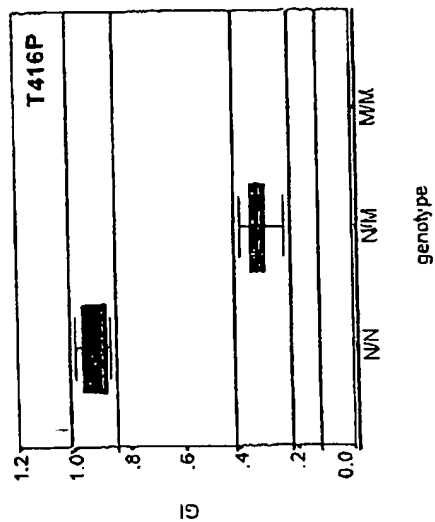


Figure 7N

# Genotype calling algorithm

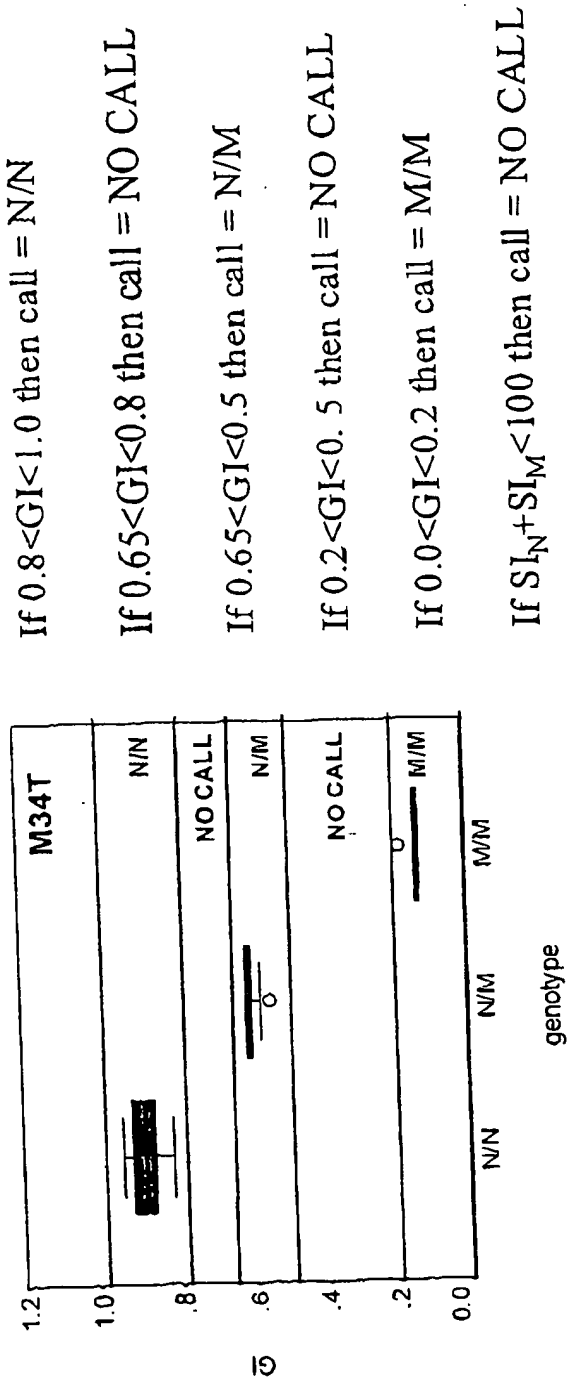
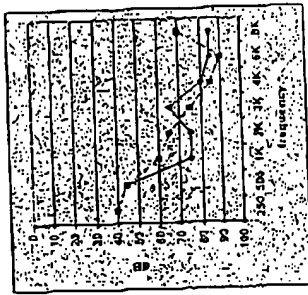


Figure 8

# Interactions between deafness genes?

- severe hearing loss
- progressive
- age of onset 10 years



Mutation	GI	Call
connexin 26		
35delG	0.992806	N/N
W24X	0.93836	N/N
M34T	0.619185	N/M
V37I	0.902981	N/N
167delT	0.999151	N/N
235delC	0.997346	N/N
L90P	0.992122	N/N
R143W	0.862635	N/N
313del14	0.932165	N/N
pendrin		
L236P	0.935414	N/N
G1001A	0.906897	N/N
E384G	0.941176	N/N
T416P	0.969543	N/N
12S rRNA		
A1555G	0.007764	M/M
usherin		
2299delG	0.995937	N/N

Figure 9

# Attachment chemistry

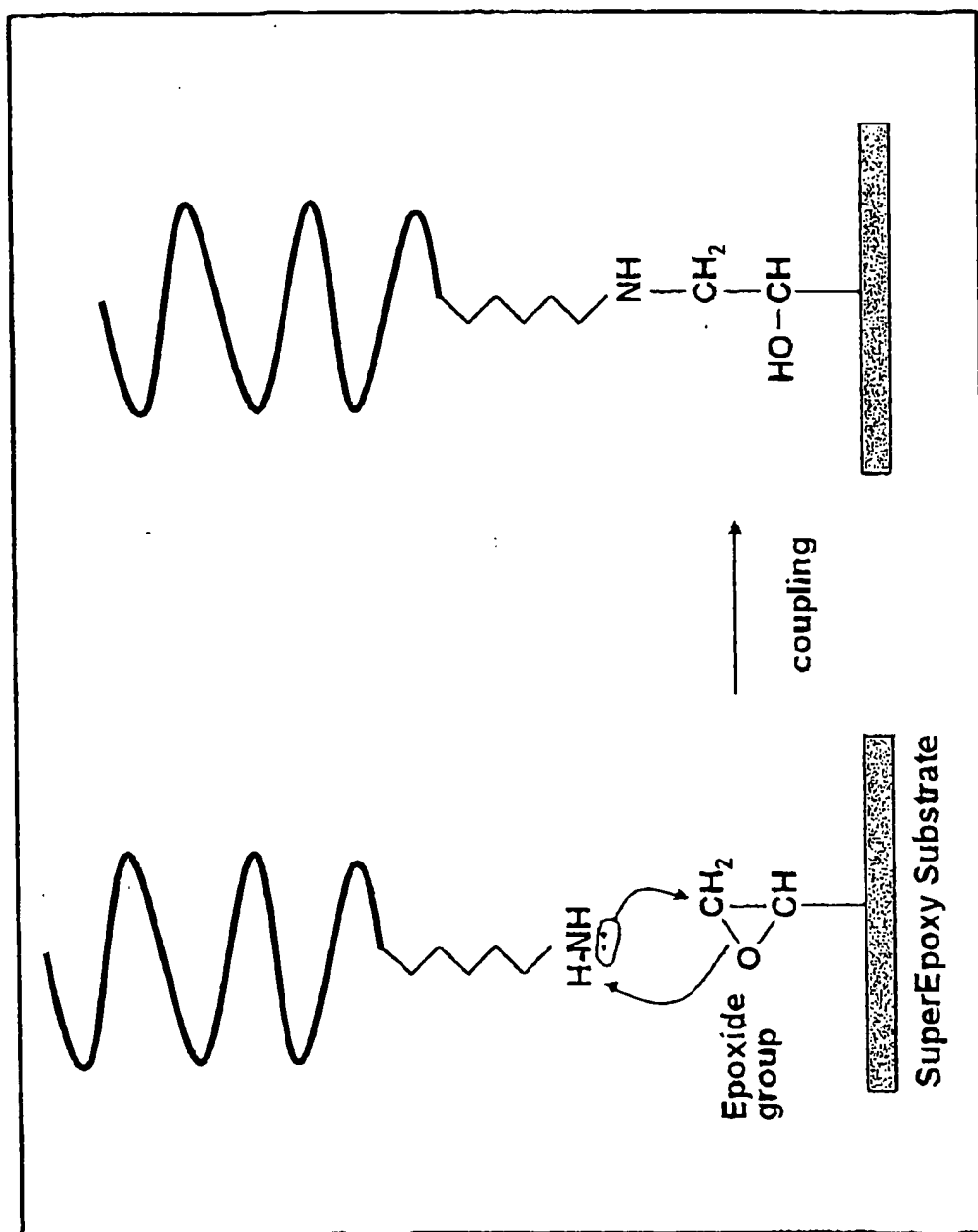


Figure 1

## Microarray based genotyping

Allele specific  
oligonucleotides (ASO)

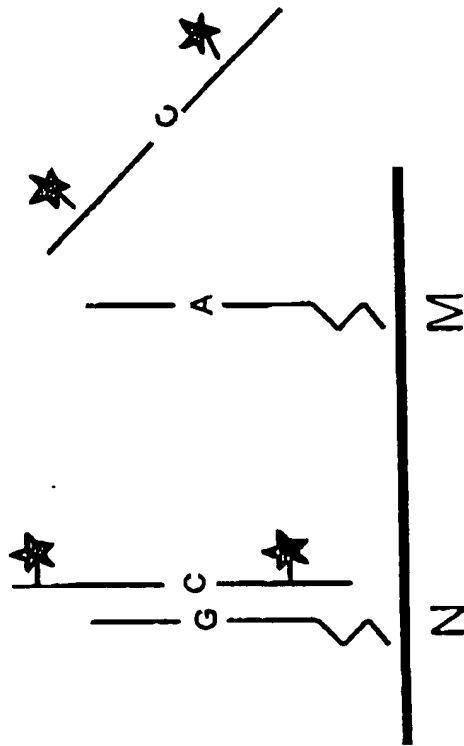


Figure 2

Step 1: Genotyping of connexin 26 35ΔG and M34T mutations (i)

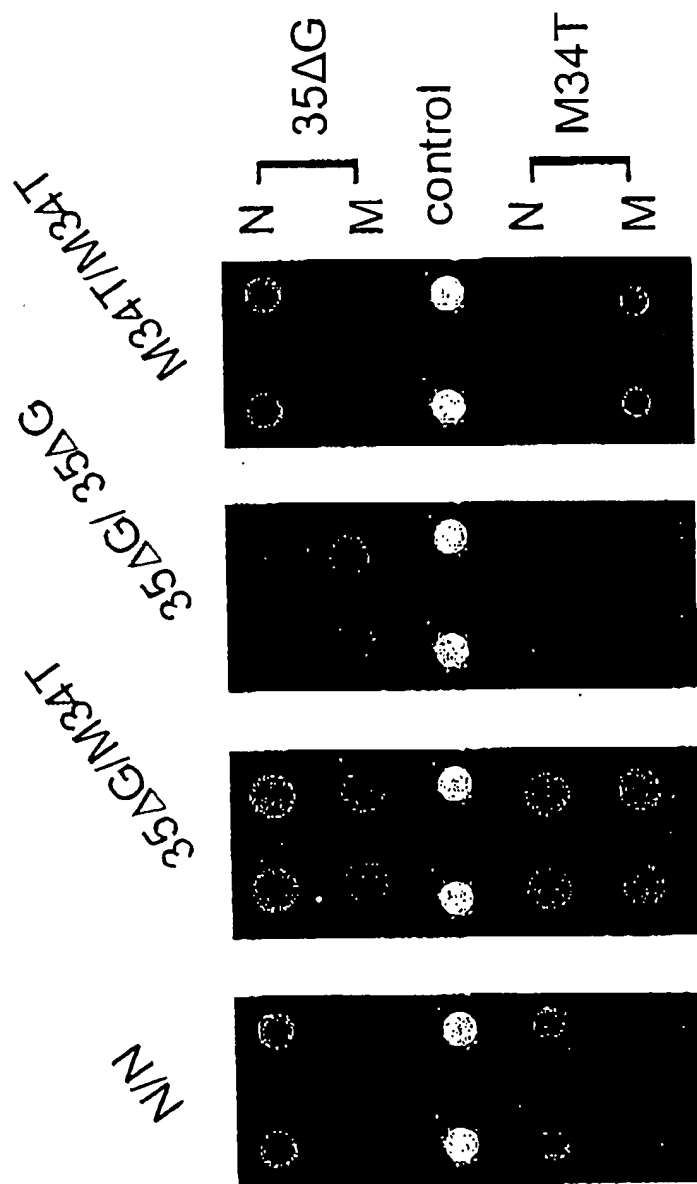


Figure 3

Step 1: Genotyping of connexin 26 35ΔG and M34T mutations (ii)

$$\text{Genotype Index (GI)} = \frac{SV_N}{(SV_N + SV_M)}$$

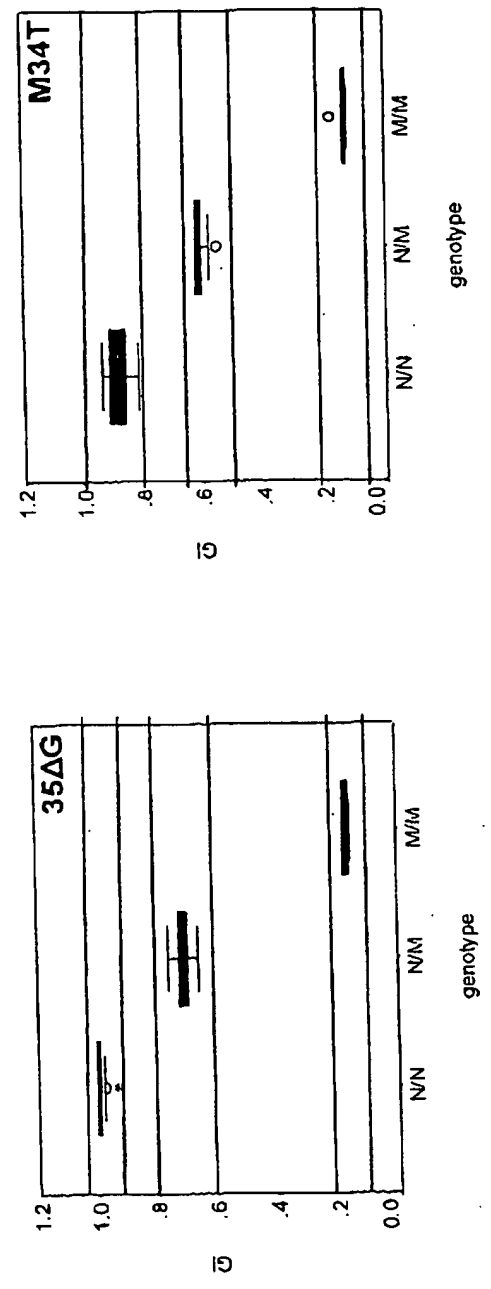


Figure 4

# Step 2: Genotyping of connexin 26 mutations

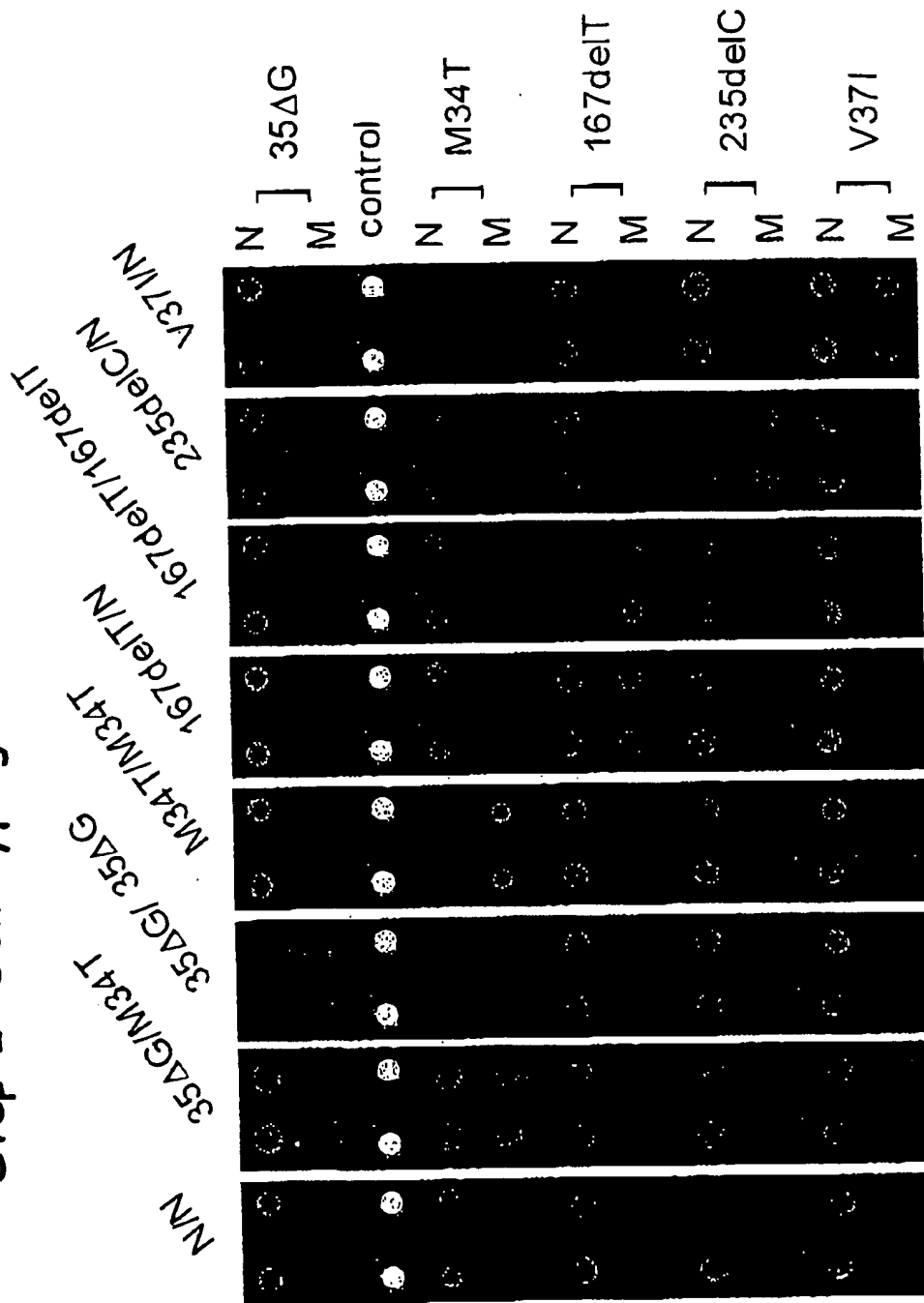


Figure 5

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20

&lt;210&gt; 62

&lt;211&gt; 20

&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; oligonucleotide 1555M21

&lt;400&gt; 62

atagaggagg caagtcgtaa

20

&lt;210&gt; 63

&lt;211&gt; 20

&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; oligonucleotide 2299W20

&lt;400&gt; 63

gggcagtgtg agtgcaaaaa

20

&lt;210&gt; 64

&lt;211&gt; 17

&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; oligonucleotide 2299M17

&lt;400&gt; 64

ggcagtgtag tgcaaaa

17

&lt;210&gt; 65

&lt;211&gt; 20

&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; primer mix 1 - P1

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&lt;212&gt; DNA

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ctgcagctga tcttcgtgtc

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&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; primer mix 3 - P7-thio

&lt;400&gt; 76

acaaagcagt ccacagtgtt

20

&lt;210&gt; 77

&lt;211&gt; 20

&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; primer mix 3 - USH2AF

&lt;400&gt; 77

atgtgagccc tgccagtgtt

20

&lt;210&gt; 78

&lt;211&gt; 21

&lt;212&gt; DNA

&lt;213&gt; artificial sequence

&lt;220&gt;

&lt;223&gt; primer mix 3 - USH2AR-thio

&lt;400&gt; 78

tcacaggcct tacaattggt g

21